# "Version with Markings to Show Changes Made" A METHOD FOR DETERMINING GENETIC AFFILIATION, SUBSTRUCTURE AND GENE FLOW WITHIN HUMAN POPULATIONS

# APR 0 1 2002 [0001]

#### **CROSS-REFERENCE**

This application claims the benefit of U.S. Provisional Application No. 06/245,355, filed November 1, 2000, which application is incorporated herein by reference.

#### STATEMENT REGARDING FEDERALLY SPONSORED RESEARCH

[0002] This invention was made with government support under grant nos. GM55273 and GM 28428 awarded by the NIH. The government may have certain rights in this invention.

#### FIELD OF THE INVENTION

[0003] The present invention relates to nucleic acid polymorphisms and their methods of use in, for example, determination of paternity and forensics.

#### BACKGROUND OF THE INVENTION

[0004] The science of genetics has taken a keen interest in the identification of human individuals and genetic relationships between individuals. The genome of an individual is unique to that individual, and can be used for identification purposes, e.g., testing for paternity and/or forensic testing (e.g. to identify an individual in the context of post-mortem identification or in the criminal justice system). Procedures have been developed which are based on identification and characterization of changes in an individual's DNA, referred to as DNA polymorphisms, where such changes are due to nucleotide substitution, insertion, or deletion within the chains of DNAs.

[0005] In forensics, for example, there is an interest in polymorphisms for identification purposes. Techniques have been developed to compare homologous segments of DNA to determine if the segments are identical or if they differ in one or more nucleotides. Practical applications of these techniques relate to fields other than forensic medicine, for example, genetic disease diagnosis and human genome mapping.

[0006] The most accurate and informative way to compare DNA segments requires a method which provides the complete nucleotide sequence for each DNA segment. Particular techniques have been developed for determining actual sequences in order to study mutation in human genes. See, for example, Proc. Natl. Acad. Sci. U.S.A. 85, 544-548 (1988) and Nature 330, 384-386 (1987). However, because of the extensive amounts of time and high costs to determine, interpret, and compare sequence information, presently it is not practical to use extensive sequencing for compare more than just a few DNA segments.

[0007] A frequently used technique for screening for DNA polymorphisms arising from mutations consist of digesting the DNA strand with restriction endonucleases and analyzing the resulting fragments by means of Southern blots. See Am. J. Hum.Genet. p32, 314-331 (1980) or Sci. Am. 258, 40-48 (1988). Since mutations often occur randomly they may affect the recognition sequence of the endonuclease and preclude the enzymatic cleavage at that site. Restriction fragment length polymorphism mappings (RFLPS) are based on changes at the restriction site. They are accurate but not very informative (PIC) 0.3). The major problem with RFLPs is the inability of a test to detect changes that do not affect cleavage with a restriction endonuclease. In addition, the methods used to detect RFLPs are very labor intensive and expensive, especially the techniques which includes Southern blot analysis.

[0008] Another technique for detecting specific mutations in particular DNA segment involves hybridizing DNA segments which are being analyzed with a complementary, labeled oligonucleotide probe. See Nucl. Acids Res. 9, 879-894 (1981). Since DNA duplexes containing even a single base pair mismatch exhibit high thermal instability, the differential melting temperature can be used to

distinguish target DNAs that are perfectly complimentary to the probe from target DNAs that only differ by a single nucleotide. See, *e.g.*, U.S. Pat. No. 4,683,194. Further, subtle genetic differences among related individuals regarding nucleotides which are substituted in the DNA chains are difficult to detect. VNTR's or Jeffrey's probes are very informative but labor intensive, in distinction to microsatellites which are equally informative PCR based tests.

Short tandem repeat (STR) polymorphisms are commonly used in DNA identification, either as adjuncts to other genetic tests, or as stand-alone tests. Typically, when STRs are used for human identification, they are amplified in groups of three to four loci (multiplex amplification). Generally, the resulting amplified fragments are analyzed by polyacrylamide gel electrophoresis. Polymorphisms are thus typed according to size by comparing to similarly labeled known external standards or differently labeled internal standards. U.S. Pat. No. 5,364,759 describes the genus of simple tandem repeats as well as a DNA typing method employing the simple tandem repeats and PCR amplification of the loci. Fragments are analyzed by differential labeling of the products.

[0010] A critical parameter in DNA typing is the power of exclusion for the system. Power of exclusion is the ability of a test to exclude a falsely accused individual based on the individual's genetic characteristics. The commonly used STR multiplexes have exclusion probabilities in the range of 85% to 91%. This compares unfavorably with restriction fragment length polymorphic loci (RFLP loci), which often provide an equivalent power with just one locus. STR testing batteries which include greater numbers of lower power systems are more susceptible to this problem than are RFLP testing batteries which include a smaller number of higher power systems. The low exclusion probabilities of commonly used STR loci are the most negative aspect of their use, although the frequencies of both alleles of an individual can be included in calculating match. Although it is simpler and faster to perform DNA typing with STR loci than with RFLP loci and it can be performed with much smaller quantities of DNA, typing using STR loci sacrifice in exclusion power. Another disadvantage of current STR multiplex DNA typing systems is that the amplification is rarely, if ever, clean. In

other words there is considerable formation of spurious bands, which is thought to be due to DNA polymerase slippage and mis-priming events (see e.g., Tautz D., Hyper variability of Simple Sequences as a General Source for Polymorphic DNA Markers, Nuc. Acids Res., 17(16) 6463-70 (1989)).

- [0011] Other polymorphisms take the form of single nucleotide variations between individuals of the same species. Such polymorphisms are far more frequent than RFLPS, STRs and VNTRs. Some single nucleotide polymorphisms occur in protein-coding sequences, in which case, one of the polymorphic forms may give rise to the expression of a defective or other variant protein and, potentially, a genetic disease. Other single nucleotide polymorphisms occur in noncoding regions. Some of these polymorphisms may also result in defective protein expression (e.g., as a result of defective splicing). Other single nucleotide polymorphisms have no phenotypic effects.
- Single nucleotide polymorphisms (SNPs) can be used in the same manner as RFLPs, and VNTRs but offer several advantages. Single nucleotide polymorphisms occur with greater frequency and are spaced more uniformly throughout the genome than other forms of polymorphism. The greater frequency and uniformity of single nucleotide polymorphisms means that there is a greater probability that such a polymorphism will be found in close proximity to a genetic locus of interest than would be the case for other polymorphisms. Also, the different forms of characterized single nucleotide polymorphisms are often easier to distinguish than other types of polymorphism, *e.g.*, by use of assays employing allele-specific hybridization probes or primers).
- [0013] There is a need in the art for a very accurate genetic relationship test procedure which uses very small amounts of an original DNA sample, yet produces very accurate results. This is particularly true in the forensic medicine area and criminology because often only very small samples of DNA available.

# SEQUENCE LISTING

[0014] The present specification incorporates herein by reference, each in its entirety, the sequence information on the Compact Disks (CDs) labeled Copy 1

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and Copy 2. The CDs are formatted on IBM-PC, with operating system compatibility with MS-Windows. The files on each of the CDS are as follows:

Copy 2 – Seglist.txt 268KB; and Copy 2 – Seglist.txt 268 KB.

#### SUMMARY OF THE INVENTION

- [0015] The present invention provides novel polymorphisms on the Y chromosome and methods of using Y chromosome polymorphisms as indicators of evolutionary heritage. The polymorphisms of particular interest in the present invention are clustered to specific regions of the Y chromosome, with polymorphisms of particular use found mostly in the Non-recombining Region of the human Y chromosome (NRY). These polymorphisms, including but not limited to SNPs, insertions, and deletions, may be useful for numerous applications, including forensics, paternity testing, diagnosis and the like.
- In one embodiment, the present invention provides nucleic acid segments of between 10 and 100 bases containing at least 10, 15 or 20 contiguous nucleotides from any of the polymorphic regions of the Y chromosome shown in TABLE 1, and may include a polymorphic site. Complements of these segments are also included. The segments can be DNA or RNA, and can be double or single-stranded. Some segments are 10-20 or 10-50 bases long and may be less than 20 or 50 bases long. Preferred nucleic acid segments allow for the identification and analysis of nucleic acid sequences on the Y chromosome which include at least one polymorphic site that is at least diallelic.
- [0017] The invention further provides allele-specific oligonucleotides that hybridize to a polymorphic region marker (M1 to M319 (excluding unassigned markers) of the Y chromosome as shown in TABLE 1, or its complement. These oligonucleotides can be probes or primers. In a particular embodiment, the nucleic acid segments include the forward and/or reverse primer sequences (e.g. primer pairs) as in Table 1. Primer pairs allow for the amplification and identification of specific polymorphic regions of the Y chromosome. Polymorphic regions of

interest for amplification and/or identification include but are not limited to the NRY regions of the Y chromosome. The polymorphic regions (polymorphic markers) shown in TABLE 1 are nucleic acids of about between 100 and 700 bases, about 200 to about 600 bases and, in some embodiments, about 250 to about 500 bases in length. Many of the polymorphic nucleic acids (polymorphic regions (markers) shown in TABLE 1 may include more than one polypmorphic site.

[0018] The invention further provides a method of analyzing a nucleic acid from an individual. The method determines which base is present at any one of the polymorphic sites of the Y chromosome as shown in TABLE 1 in bold type. Optionally, a set of bases occupying a set of the polymorphic sites shown in TABLE 1 is determined. This type of analysis can be performed on a plurality of individuals who are tested for the presence of a particular polymorphism by identifying specific polymorphic markers. The polymorphism can be correlated with a base or set of bases present at the polymorphic sites in the individuals tested, and the evolutionary heritage of the individual can be indicated by the presence or absence of a particular polymorphism.

In one embodiment, the invention provides a method for determining the ethnic origin of a male, comprising obtaining a nucleic acid sample from the male and identifying at least two polymorphic markers in the nucleic acid sample indicative of the ethnic origin of the male, using at least one primer pair from TABLE 1. The identifying of the polymorphic markers may indicate the ethnic origin of the male as being at least one of the haplotype groups selected from the group consisting of haplotype Group I, Group II, Group III, Group IV, Group V, Group VI, Group VIII, Group IX or Group X. In some embodiments, at least one polymorphic marker identified is a polymorphic marker from TABLE 1. The polymorphic markers may identify a haplotype associated with a haplotype group selected from the group consisting of haplotype Group I, Group II, Group III, Group IV, Group V, Group VI, Group VIII, Group IX or Group X, or a sub-haplotype group for the ethnic origin of the male.

- [0020] In another embodiment, the invention provides a method for identifying a plurality of polymorphic sites in a nucleic acid, comprising obtaining a sample of the nucleic acid from at least one individual, and identifying, in the nucleic acid, at least one of the polymorphic sites in at least two polymorphic markers of TABLE 1. The sample of nucleic acids may be obtained from a plurality of individuals, with the presence of the polymorphic markers in each sample of the nucleic acid determined for each of the individuals. The method may further comprise testing each individual for presence of a group of polymorphic markers which identify the haplotype of each individual, wherein the haplotype is indicative of a geographic distribution of a population or an ancestral population.
- [0021] In still other embodiments, the invention provides a method for determining the ethnic origin of a human male individual, comprising obtaining a nucleic acid sample from the male, testing the nucleic acid sample for presence of a plurality of polymorphic markers selected from TABLE 1, identifying which polymorphic markers are present in the nucleic acid sample, and assigning a haplotype group to the male based on the identified markers, wherein the haplotype group is indicative of the ethnic origin of the male.
- [0022] In certain embodiments, the invention provides a method for determining the paternity of a human male individual, comprising obtaining a nucleic acid sample from the male, testing the nucleic acid sample for the presence of a plurality of polymorphic markers from TABLE 1, identifying which polymorphic markers are present in the nucleic acid sample, and comparing the identified polymorphic markers to a set of polymorphic markers identified in nucleic acid samples from potential fathers.
- [0023] The invention additionally provides a kit for determining ethnic origin of an individual, comprising at least two primer pairs capable of identifying at least two polymorphic markers from TABLE 1. The kit may further comprise a control

nucleic acid for detecting the presence or absence of the polymorphic markers from TABLE 1.

- [0024] The invention further comprises a set of primers and enzymes useful in performing an assay to identify particular polymorphisms in human male DNA. A method of identifying polymorphisms is disclosed whereby a sample is provided and subjected to amplification using primers of the invention and thereafter determining sequences (polymorphic regions) which were amplified.
- [0025] A feature of the invention is that polymorphisms not previously identified are described herein, and are associated with a particular haplotype, indicative of a specific evolutionary heritage.
- [0026] An advantage of the invention is that the sequences disclosed herein can be used in a range of different assay systems to determine the presence of a polymorphism in a sample.
- [0027] A feature of the invention is a method for analyzing a set of unique polymorphisms on the Y chromosome to determine and identify an individual's evolutionary heritage and/or ethnicity.
- [0028] A feature of the invention is to provide a kit for determining an individual's geographical or ethnic origins.
- [0029] These and other objects, advantages, and features of the invention will become apparent to those persons skilled in the art upon reading the details of the invention as fully described below.

# BRIEF DESCRIPTION OF THE DRAWINGS

- [0030] Fig. 1. Contemporary worldwide distribution of Y chromosome groups in 22 regions determined by the methods and compositions of the invention.
- [0031] Fig. 2. A phylogenetic tree deduced from 167 NRY polymorphisms on the principle of maximum parsimony.

- [0032] Fig. 3. Maximum likelihood network inferred from the haplotype frequencies.
- [0033] Fig. 4. Maximum parsimony phylogeny of human NRY chromosome biallelic variation.

#### DETAILED DESCRIPTION OF PREFERRED EMBODIMENTS

- [0034] Before the present polymorphisms and detection methods are described, it is to be understood that this invention is not limited to particular methods or polymorphisms described, as such may, of course, vary. It is also to be understood that the terminology used herein is for the purpose of describing particular embodiments only, and is not intended to be limiting, since the scope of the present invention will be limited only by the appended claims.
- Where a range of values is provided, it is understood that each intervening value, to the tenth of the unit of the lower limit unless the context clearly dictates otherwise, between the upper and lower limits of that range is also specifically disclosed. Each smaller range between any stated value or intervening value in a stated range and any other stated or intervening value in that stated range is encompassed within the invention. The upper and lower limits of these smaller ranges may independently be included or excluded in the range, and each range where either, neither or both limits are included in the smaller ranges is also encompassed within the invention, subject to any specifically excluded limit in the stated range. Where the stated range includes one or both of the limits, ranges excluding either or both of those included limits are also included in the invention.
- [0036] Unless defined otherwise, all technical and scientific terms used herein have the same meaning as commonly understood by one of ordinary skill in the art to which this invention belongs. Although any methods and materials similar or equivalent to those described herein can be used in the practice or testing of the present invention, the preferred methods and materials are now described. All

publications mentioned herein are incorporated herein by reference to disclose and describe the methods and/or materials in connection with which the publications are cited.

[0037] It must be noted that as used herein and in the appended claims, the singular forms "a", "and", and "the" include plural referents unless the context clearly dictates otherwise. Thus, for example, reference to "a nucleic acid" includes a plurality of such nucleic acids and reference to "the primer" includes reference to one or more primers and equivalents thereof known to those skilled in the art, and so forth.

[0038] The publications discussed herein are provided solely for their disclosure prior to the filing date of the present application. Nothing herein is to be construed as an admission that the present invention is not entitled to antedate such publication by virtue of prior invention. Further, the dates of publication provided may be different from the actual publication dates which may need to be independently confirmed.

#### THE INVENTION IN GENERAL

[0039] The use of certain nucleotide repeat polymorphisms for identifying or comparing DNA segments have been described. (See *e.g.*, Weber & May *Am Hum Genet* 44:388 (1989), Litt & Luthy *Am Hum Genet* 44:397(1989)). The present invention is based on the finding that particular polymorphisms on the Y chromosome, including the novel polymorphisms included herein, are indicative of the evolutionary heritage and/or a paternal lineage in an individual having a Y chromosome (e.g., a male or XXY individual). These particular polymorphic genetic segments, and primers used to identify the polymorphisms for identification and comparison purposes, correspond to regions of the Y chromosome having clustered polymorphisms that are homopolymeric in regions which exhibit a very low mutation rate. An advantage of the polymorphisms of the invention is that no recombination occurs in the regions containing these markers, and thus the accumulation of mutations is preserved as an intact

haplotype. This creates a genetic profile that remains intact across the generations. If men share the same derived allele, then they are identical by descent, not just by state. While a very small amount of recurrent or revertant back mutation has been observed at some markers, these anomalies are easily recognized as such because of the high resolution of the Y tree. The recognition of new Y-chromosome markers represents a major leap in the investigation of human genetic diversity (in male lineages, complementing the information from female lineages derived from mitochondrial DNA).

[0040] The polymorphisms and methods of the present invention provide a simple way of identifying male siblingship as well as a genetic route to identify male children by so called "genebanking" using DNA or blood, or saliva from a child. Also the Y chromosome polymorphisms can reveal patterns (estimates) of recent gene flow from one gene pool to another, i.e. admixture. The methods of the present invention make the large amount of information contained in the phylogeny of haplotypes accessible for analysis.

# **DEFINITIONS**

[0041] The term "oligonucleotide" as used herein can be DNA, RNA, or a substituted variation of these nucleic acids. The oligonucleotide may be single- or double-stranded. Oligonucleotides can be naturally occurring or synthetic, but are typically prepared by synthetic means. Preferred oligonucleotides of the invention include segments of DNA, or their complements including any one of the polymorphic sites shown in TABLE 1. The segments are usually between 5 and 100 bases (nucleotides), and often between 5-10, 5-20, 10-20, 10-50, 20-50 or 20-100 bases. The polymorphic site can occur within any position of the segment. The segments can be from any of the allelic forms of DNA shown in TABLE 1.

[0042] The term "hybridization probes" as used herein refers to oligonucleotides capable of binding in a base-specific manner to a complementary strand of nucleic acid. Such probes include peptide nucleic acids, as described in Nielsen et al., Science 254, 1497-1500 (1991).

least a single-stranded portion that is adapted to act as a point of initiation of template-directed DNA synthesis under appropriate conditions (i.e., in the presence of four different nucleoside triphosphates and an agent for polymerization, such as, DNA or RNA polymerase or reverse transcriptase) in an appropriate buffer and at a suitable temperature. The appropriate length of a primer depends on the intended use of the primer but typically ranges from 15 to 30 nucleotides. A primer need not reflect the exact sequence of the template but must be sufficiently complementary to hybridize with a template.

[0044] The term "primer site" as used herein refers to the area of the target DNA to which a primer hybridizes. The term "primer pair" as used herein refers to a set of primers including at least one 5' upstream primer that hybridizes with the 5' end of the DNA sequence to be amplified (a forward or "for" primer) and at least one 3' downstream primer that hybridizes with the complement of the 3' end of the sequence to be amplified (a reverse or "rev" primer). Primer pairs allow for the amplification and identification of corresponding polymorphic regions.

[0045] The term "polymorphic site" is used herein to describe mutations within a nucleic acid sequence which include but are not limited to site specific mutations, insertions and deletions, these mutations being found in the nucleic acid of some individuals and not in others, e.g. the polymorphic site identifies a specific polymorphism of an individual. The present invention provides segments of nucleic acid which contain at least one polymorphic site (i.e. polymorphic region). These "polymorphic regions" of the Y chromosome can be analyzed to identify a specific polymorphic site which in turn identifies a specific polymorphism associated with certain individuals.

[0046] The polymorphic regions of the present invention are also defined as "polymorphic markers" due to their usefulness in marking (identifying specific polymorphic sites). The polymorphic markers of the present invention identify specific haplotypes in the male population, these haplotypes being indicative of a specific geographical or ethnic origin. Certain polymorphic markers which identify a polymorphism shared by a large group of individuals allow for the

grouping of those haplotypes which share that marker. These more commonly found markers are found at the branch points of a phylogenetic tree and are crucial in separating individuals into unique haplotype groups. The haplotype groups have this ancestral marker which branches off from a point earlier in the phylogenetic tree. The polymorphic markers of the present invention have identified over 171 haplotypes which can be divided into ten haplotype groups.

[0047] The term "polymorphism" as used herein refers to the occurrence of two or more genetically determined alternative sequences or alleles in a population. A polymorphic marker or site is the locus at which divergence occurs. Preferred markers have at least two alleles, each occurring at a frequency of greater than 1%, and more preferably greater than 10% or 20% of a selected population, and can be present at a frequency greater than 30% to 50% or more in selected portions of the population. A polymorphic locus may be as small as one base pair. Polymorphic markers include restriction fragment length polymorphisms, VNTR's, hypervariable regions, minisatellites, dinucleotide repeats, trinucleotide repeats, tetranucleotide repeats, simple sequence repeats, and insertion elements such as Alu. The first identified allelic form is arbitrarily designated as the reference form and other allelic forms are designated as alternative or variant alleles. Polymorphisms refer to sequence differences between a reference form and a selected allele, and encompasses single or multiple nucleotide differences which can result from nucleotide insertion(s), deletion(s), substitution(s) and/ or a combination thereof. The allelic form occurring most frequently in a selected population is sometimes referred to as the wildtype form. Diploid organisms may be homozygous or heterozygous for allelic forms. A diallelic polymorphism has two forms. A triallelic polymorphism has three forms. The term "polymorphism" as used herein refers to any detectable polymorphic site in DNA or RNA that is detectable using the present methods. The term as used herein encompasses, for example, polymorphisms associated with a disease state (i.e. mutations), "silent" polymorphisms (i.e. associated with a wild-type phenotype or in a non-coding

region), and polymorphisms associated with a predisposition and/or response to treatment (i.e. a polymorphism in an allele of a gene).

interchangeably herein refers to a polymorphism" and "SNP" as used interchangeably herein refers to a polymorphic site occupied by a single nucleotide (i.e. single base), which is the site of variation between allelic sequences. In general, SNPs are DNA sequence variations that occur when a single nucleotide (A, T, C or G) in the genomic sequence is altered. For example a SNP might change the DNA sequence AAGGCTAA to ATGGCTAA. SNPs can occur in both coding (gene) and noncoding regions of the genome. The site is usually preceded by and followed by highly conserved sequences of the allele (e.g., sequences that vary in less than 1/100 or 1/1000 members of the population).

[0049] A single nucleotide polymorphism usually arises due to substitution of one nucleotide for another at the polymorphic site. A transition is the replacement of one purine by another purine or one pyrimidine by another pyrimidine. A transversion is the replacement of a purine by a pyrimidine or vice versa. Single nucleotide polymorphisms can also arise from a deletion of a nucleotide or an insertion of a nucleotide relative to a reference allele. Hybridizations are usually performed under stringent conditions, for example, at a salt concentration of no more than 1M and a temperature of at least 25°C. For example, conditions of 5X SSPE (750 mM NaCl, 50 mM NaPhosphate, 5 mM EDTA, pH 7.4) and a temperature of 25°-30°C are suitable for allele-specific probe hybridizations.

[0050] The term "isolated nucleic acid" as used herein refers to a nucleic acid isolated from an individual that is the predominant species present (i.e., on a molar basis it is more abundant than any other individual species in the composition). Preferably, an isolated nucleic acid comprises at least about 50, 80 or 90 percent (on a molar basis) of all macromolecular species present. Most preferably, the object species is purified to essential homogeneity, i.e. contaminant species cannot be detected in the composition by conventional detection methods. The isolated nucleic acid includes a selected DNA fragment (e.g., isolated by an amplification reaction), and an isolated mRNA.

[0051] The term "evolutionary heritage" as used herein refers to the association of a particular polymorphism with a population having a particular geographic distribution. This includes polymorphisms that are indicative of an ancestral population, i.e. a population from which an individual is a descendant.

#### GENERAL ASPECTS OF THE INVENTION

- [0052] The present application provides novel polymorphisms, including polymorphisms clustered in and around a non-recombining portion of the human Y chromosome (NRY) The polymorphic sites and the regions flanking these polymorphic sites are shown in TABLE 1.
- [0053] By knowing sequences which include particular polymorphisms on the Y chromosome, primers based on these sequences can be used in detection assays. The primers can be provided in assay kits which cover from one to any and all of the polymorphisms developed here and the kits may further comprise appropriate enzymes for use with the primers and/or reagents for the isolation and processing of nucleic acids from an individual.
- [0054] The methods and compositions of the present invention allow for the genetic typing of male individuals into ten major haplotype groups. The markers and primer sets shown in TABLE 1 allow not only for typing males into one of the haplotype groups or a combination of haplotype groups, but also enables an individual to be identified to a specific geographical area associated with haplotype group. Figure 1 shows a contemporary worldwide frequency distribution of the 10 Y chromosome groups in 22 regions. Each group is represented by a distinguishing color. Colored sectors reflect representative group frequencies. The frequency distribution of the ten groups is based on > 1000 globally diverse samples genotyped using a hierarchical top down approach as illustrated in FIG.1 above the global map. The representative branching and frequency of polymorphic markers in TABLE 1 are also shown in FIG. 1 (individual marker numbers are not shown).

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[0055] The identification of an individuals haplotype is base on identifying the presence of at least two distinct polymorphic markers (i.e. at least two distinct polymorphic sites must be identified), for example, polymorphic markers M91 and M278 identify haplotype 9(shown in FIG. 2 and FIG. 4). More likely, determining the haplotype of an individual involves the identification of 3 or more markers, usually at least about 3 to 7 markers, or 7 to 9 markers or even 9 or more markers.

Haplotype groups comprise haplotypes which have at least one ancestral marker which branches off from a point earlier in the phylogenetic tree. For example, marker 91 (M91) identifies haplotypes in Group I while haplotypes in group V are identified by one marker from each of the following sets of markers; one marker from {M42, M94, M139, M251, M299} plus one from {M168, M294} and one marker from {RPS4Y, M216, M316}. To determine which haplotype group and individual is associated with, the individuals nucleic acid would need to be analyzed with at least eleven polymorphic markers. For exemplary purposes, an individuals nucleic acid could be assayed for the presence and absence of the following markers; M91, M299, M249, M294, M203, M96, M316, M9, M74, M207, M214 to determine which haplotype group they are associated with which is indicative of a certain geographical or ethnic origin.

Fig. 1 illustrates that haplotype Group I is mainly associated with Africa and in particular, southern and eastern Africa (approximately about 90% of males of haplotype Group I are of African origin). Haplotype Groups II (about 80% to about 99% frequency distribution (f.d.)) and III (about 75% to about 95% f.d.) are also strongly related to Africa compared to Groups IV through X. Populations represented in Groups I and II include some Khoisan and Bantu speakers from South Africa, Pygmies from central Africa, and lineages in Sudan, Ethiopia and Mali. Virtually all men with Group I and II haplotypes are of African affiliation from a paternal perspective. Group III lineages are predominantly African, although a sub-set of Group III lineages occur in populations bordering the Mediterranean (Middle East, Turkey, North Africa, Southern Europe).

[0058] Approximately about 70% to about 99% of the males in Group IV are of Japanese origin. Group V is slightly associated with Japan (about 10% to about 25% f.d.) and Indonesia (about 10% to about 35% frequency) with the largest frequency being associated with Australia and central Asians (about 45% to about 75% f.d.).

[0059] Group VI is more widely distributed than other haplotypes, covering the geographical area of Europe, Eastern Europe, Asia, and India. The presence of haplotype group VI in North America, Australia and Polynesia is a consequence of recent human movements since C. Columbus catalyzed the age of exploration. The largest Group VI frequency is associated with southern Europe and the middle east, with a distribution frequency of about 60% to about 85%.

[0060] Group VII is more widely associated with eastern Asia and Indonesia with distribution frequencies ranging from about 75% to about 99%. Group VIII is almost exclusively found in Papua-New Guniea (distribution frequencies of about 70% to about 95%) with a slight distribution in central Asia (distribution frequency of about 1% to about 30%). Recently, there is evidence which indicates the presence of Group VIII in Indonesia. Other specific Group VIII lineages occur in India and Europe. Individuals of haplotype Group IX are mostly associated Europe (about 75% to about 95% f.d.), India (about 25% to about 50% f.d.). Their occurrence in North America (about 35% to about 55%) Australia (35%), Polynesia is a consequence of European gene flow during the last 500 years.

[0061] Group X individuals are geographically associated with Central Asia and the Americas with a frequency distribution in North America of about 25% to about 50%, Central America of about 75% to about 95% and in South America of about 80% to about 99%. The above distribution frequencies of the various haplotypes in the geographic regions mentioned above are only representative ranges of the haplotype frequencies worldwide.

# **Analysis of Polymorphisms**

[0062] Polymorphisms are detected in a target nucleic acid from an individual being analyzed. For assay of genomic DNA, virtually any biological sample (other than pure red blood cells) is suitable. For example, convenient tissue samples include whole blood, semen, saliva, tears, urine, fecal material, sweat, buccal, skin and hair. For assay of cDNA or mRNA, the tissue sample must be obtained from an organ in which the target nucleic acid is expressed. For purposes of the present invention, the sample is obtained from a male, and preferably a human male.

[0063] Many of the methods described below require amplification of DNA from target samples. This can be accomplished by *e.g.*, PCR. See generally PCR Technology: Principles and Applications for DNA Amplification (ed. H. A. Erlich, Freeman Press, N.Y., N.Y., 1992); PCR Protocols: A Guide to Methods and Applications (eds. Innis, et al., Academic Press, San Diego, Calif., 1990); Mattila et al., Nucleic Acids Res. 19, 4967 (1991); Eckert et al., PCR Methods and Applications 1, 17 (1991); PCR (eds. McPherson et al., IRL Press, Oxford); and U.S. Pat. No. 4,683,202.

[0064] Other suitable amplification methods include the ligase chain reaction (LCR) (see Wu and Wallace, Genomics 4, 560 (1989), Landegren et al., Science 241, 1077 (1988), transcription amplification (Kwoh et al., Proc. Natl. Acad. Sci. USA 86, 1173 (1989)), and self-sustained sequence replication (Guatelli et al., Proc. Nat. Acad. Sci. USA, 87, 1874 (1990)) and nucleic acid based sequence amplification (NASBA). The latter two amplification methods involve isothermal reactions based on isothermal transcription, which produce both single stranded RNA (ssRNA) and double stranded DNA (dsDNA) as the amplification products in a ratio of about 30 or 100 to 1, respectively.

# Detection of Polymorphisms in Target DNA

[0065] There are two distinct types of analysis depending whether a polymorphism in question has already been characterized. The first type of analysis is sometimes referred to as *de novo* characterization. This analysis

compares target sequences in different individuals to identify points of variation, e.g., polymorphic sites, SNPs. By analyzing groups of individuals representing the greatest ethnic diversity among humans and greatest breed and species variety in plants and animals, patterns characteristic of the most common alleles/haplotypes of the locus can be identified, and the frequencies of such populations in the population determined. Additional allelic frequencies can be determined for subpopulations characterized by criteria such as geographical distribution and ancestral ethnicity. The *de novo* identification of the polymorphisms of the invention is described in the Examples section. The second type of analysis is determining which form(s) of a characterized polymorphism are present in individuals under test. There are a variety of suitable procedures, which are discussed in turn.

# Allele-Specific Probes

[0066]

The design and use of allele-specific probes for analyzing polymorphisms is described by e.g., Saiki et al., Nature 324, 163-166 (1986); Dattagupta, EP 235,726, Saiki, WO 89/11548. Allele-specific probes can be designed that hybridize to a segment of target DNA from one individual but do not hybridize to the corresponding segment from another individual due to the presence of different polymorphic forms in the respective segments from the two individuals. Hybridization conditions should be sufficiently stringent that there is a significant difference in hybridization intensity between alleles, and preferably an essentially binary response, whereby a probe hybridizes to only one of the alleles. Probes with such specificity allow for the determination of a specific base occupying a polymorphic site in a sequence of a polymorphic region. Some probes are designed to hybridize to a segment of target DNA such that the polymorphic site aligns with a central position (e.g., in a 15 mer at the 7 position; in a 16 mer, at either the 8 or 9 position) of the probe. This design of probe achieves good discrimination in hybridization between different allelic forms.

[0067] Allele-specific probes are often used in pairs, one member of a pair showing a perfect match to a reference form of a target sequence and the other

member showing a perfect match to a variant form. Several pairs of probes can then be immobilized on the same support for simultaneous analysis of multiple polymorphisms within the same target sequence.

# Tiling Arrays

The polymorphisms can also be identified by hybridization to nucleic acid arrays, some example of which are described by WO 95/11995. The same array or a different array can be used for analysis of characterized polymorphisms. WO 95/11995 also describes subarrays that are optimized for detection of a variant form of a precharacterized polymorphism. Such a subarray contains probes designed to be complementary to a second reference sequence, which is an allelic variant of the first reference sequence. The second group of probes is designed by the same principles as described in the Examples except that the probes exhibit complementarily to the second reference sequence. The inclusion of a second group (or further groups) can be particular useful for analyzing short subsequences of the primary reference sequence in which multiple mutations are expected to occur within a short distance commensurate with the length of the probes (i.e., two or more mutations within 9 to 21 bases).

#### Allele-Specific Primers

[0069] An allele-specific primer hybridizes to a site on target DNA overlapping a polymorphism and only primes amplification of an allelic form to which the primer exhibits perfect complementarily. See Gibbs, Nucleic Acid Res. 17, 2427-2448 (1989). This primer is used in conjunction with a second primer which hybridizes at a distal site. Amplification proceeds from the two primers leading to a detectable product signifying the particular allelic form is present. A control is usually performed with a second pair of primers, one of which shows a single base mismatch at the polymorphic site and the other of which exhibits perfect complementarily to a distal site. The single-base mismatch prevents amplification and no detectable product is formed. The method works best when the mismatch is included in the 3'-most position of the oligonucleotide aligned with the

polymorphism because this position is most destabilizing to elongation from the primer. See, e.g., WO 93/22456.

#### Direct-Sequencing

[0070] The direct analysis of the sequence of polymorphisms of the present invention can be accomplished using either the dideoxy chain termination method or the Maxam Gilbert method (see Sambrook et al., Molecular Cloning, A Laboratory Manual (2<sup>nd</sup> Ed., CSHP, New York 1989); Zyskind et al., Recombinant DNA Laboratory Manual, (Acad. Press, 1988)). In a preferred embodiment, the direct sequencing would be carried using fluorescent sequencing, *e.g.*, using a PE Biosystems 373A sequencer.

# Denaturing Gradient Gel Electrophoresis

[0071] Amplification products generated using the polymerase chain reaction can be analyzed by the use of denaturing gradient gel electrophoresis. Different alleles can be identified based on the different sequence-dependent melting properties and electrophoretic migration of DNA in solution. Erlich, ed., PCR Technology, Principles and Applications for DNA Amplification, (W.H. Freeman and Co, New York, 1992), Chapter 7.

#### Single-Strand Conformation Polymorphism Analysis

[0072] Alleles of target sequences can be differentiated using single-strand conformation polymorphism analysis, which identifies base differences by alteration in electrophoretic migration of single stranded PCR products, as described in Orita et al., Proc. Nat. Acad. Sci. 86, 2766-2770 (1989). Amplified PCR products can be generated as described above, and heated or otherwise denatured, to form single stranded amplification products. Single-stranded nucleic acids may refold or form secondary structures which are partially dependent on the base sequence. The different electrophoretic mobilities of single-stranded amplification products can be related to base-sequence difference between alleles of target sequences.

#### **Detection of SNP Polymorphisms**

[0073] Where the polymorphism is a SNP, any suitable method known in the art can be used in their detection. For example, the present methods can utilize the detection of SNPs by DHPLC (see U.S. Pat. No. 5,795,976) to isolate and analyze specific SNPs on the Y chromosome of a large number of individuals in a fast, efficient and inexpensive manner. This method involves separating heteroduplex and homoduplex nucleic acid molecules (e.g., DNA or RNA) in a mixture using high performance liquid chromatography under partially denaturing conditions. In a preferred embodiment, the SNPs are identified on the Y chromosome using techniques such as those disclosed in co-pending application US Application Serial No. 09/502,558, February 10, 2000.

#### Mass Spectrometry

- [0074] Mass spectrometry can also be used in the methods of the present invention to verify a polymorphism and/or to identify additional polymorphisms. The mass spectrum of a nucleic acid containing the polymorphic site can be compared to the mass spectrum of nucleic acids obtained from samples of known residues at the polymorphic site. These known spectra are referred to as "signature" spectra. A simple comparison of the sample spectrum vs. signature spectra will reveal whether an individual's DNA has a specific base occupying the polymorphic site. Although sequencing of fragments of nucleic acids is possible using mass spectrometry, actual sequencing of the nucleic acid is not required for this mutational analysis. Less preparation and analysis is needed to prepare and analyze a complete, intact fragment as compared to treating a sample for actual sequencing.
- [0075] Certain mass spectrometry techniques can be used to analyze for polymorphisms. Short oligomers, *e.g.*, from one nucleotide up to approximately 50 nucleotides, can be analyzed and the resulting spectra compared with signature spectra of samples known to be wild-type or to contain a known polymorphism. A comparison of the locations (mass) and heights (relative amounts) of peaks in the

sample with the known signature spectra indicate what type of polymorphism, if any, is present. Exemplary protocols are described in U.S. Pat Nos. 5,872,003, 5,869,242, 5,851,765, 5,622,824, and 5,605, 798, which are incorporated herein by reference for teaching such techniques.

[0076] After determining polymorphic form(s) present in an individual at one or more polymorphic site on the Y chromosome, this information can be used in a number of methods.

#### Methods of Use of the Polymorphisms of the Invention

The methods of the invention have utility in a wide variety of fields where it is desirable to identify known polymorphisms of a particular individual and/or to determine allelic distribution in a group or population. Such methods include, but are not limited to, linkage analysis for the identification of disease loci, evolutionary studies to determine rates of evolution in a population, identification of polymorphisms useful in forensic identification, identification of mutations associated with a disease or predisposition, genetic marker development, and the like.

#### **Forensics**

[0078] Determination of which polymorphic sites an individual possesses, identifies a haplotype, which refers to a set of polymorphic markers that distinguishes the individual. See generally National Research Council, *The Evaluation of Forensic DNA Evidence* (Eds. Pollard et al., National Academy Press, DC, 1996). Since the polymorphic sites of the invention are generally within a region of about 50,000 bp in the human genome, the probability of recombination between these polymorphic sites is low. The more sites that are analyzed the lower the probability that the set of polymorphic markers for one individual is the same as that in an unrelated individual. If multiple polymorphic sites are analyzed, the sites are usually in different polymorphic regions (on different polymorphic markers). Thus, polymorphisms of the invention may be

used in conjunction with polymorphisms in distal genes. Preferred polymorphisms for use in forensics are diallelic because the population frequencies of two polymorphic forms can usually be determined with greater accuracy than those of multiple polymorphic forms at multi-allelic loci.

[0079] An exemplary set of polymorphic markers useful for identifying the haplotype group of an individual are the following; Markers 304(Group VI, Mediterranean), 242 (Group X, C. Asia, India, Americas), 269 (Group IX, W. Europe), 207 (Group IX, Europe, W. Asia), 74 (Groups IX-X, global), 214 (Group VII, E. Asia), 9 (Groups VII-X, global), 235 (Groups VI-X, global), 316 (Group V, Asia, America, Polynesia, Melanesia), 174 (Group IV, Asia, Japan), 299 (Groups II-X, global), 246 (Group I, Africa), 249 (Group II, Africa) 294 (Groups III-X, global), 96 (Group III, Africa, Mediterranean).

[0800] The capacity to identify a distinguishing or unique set of forensic markers in an individual is useful for forensic analysis. For example, one can determine whether a blood sample from a suspect matches a blood or other tissue sample from a crime scene by determining whether the set of polymorphic forms occupying selected polymorphic sites is the same in the suspect and the sample. If the set of polymorphic markers does not match between a suspect and a sample, it can be concluded (barring experimental error) that the suspect was not the source of the sample. If the set of markers does match, one can conclude that the DNA from the suspect is consistent with that found at the crime scene. If frequencies of the polymorphic forms at the loci tested have been determined (e.g., by analysis of a suitable population of individuals), one can perform a statistical analysis to determine the probability that a match of suspect and crime scene sample would occur by chance. If several polymorphic loci are tested, the cumulative probability of non-identity for random individuals becomes very high (e.g., one billion to one). Such probabilities can be taken into account together with other evidence in determining the innocence or guilt of an individual suspected of a criminal act.

[0081] The polymorphisms of the present invention are especially useful in identifying samples having genetic material from multiple individuals, since the

polymorphisms are single copy. Thus, the detection of more than one polymorphic Y chromosome allele in a single sample is indicative of the presence of nucleic acids from multiple individuals within the sample. Such information can be useful, for example, when multiple perpetrators are suspected of participating in a crime, or in the case of mixed unidentified remains at a grave site or accident scene.

[0082] The polymorphic sites and methods of the present invention are also useful in categorizing victims of violent crimes into ethnic and geographical groups. When a large number of victims need to be identified at a crime site, categorizing recovered victims by ethnicity can decrease the overall time for victim identification by reducing the number of comparison samples (samples from members of the victims family) to those of similar geographical origin.

# Paternity Testing

the father of a child. In most cases, the mother of the child is known and thus, the mother's contribution to the child's genotype can be traced. Paternity testing investigates whether the part of the child's genotype not attributable to the mother is consistent with that of the putative father. Paternity testing can be performed by analyzing sets of polymorphisms (polymorphic markers) in the putative father and the child. The polymorphic markers of the present invention can be useful in determining paternity of a male child, as they are specific to the Y chromosome. The mother need not be tested in such a case, as the mother has no contribution to the child's genotype as it pertains to the Y chromosome.

[0084] If the set of polymorphisms in the child attributable to the father does not match the putative father, it can be concluded, barring experimental error, that the putative father is not the real father. If the set of polymorphisms in the child attributable to the father does match the set of polymorphisms of the putative father, a statistical calculation can be performed to determine the probability of coincidental match. An exemplary method of determining the probability of parentage exclusion, i.e. the probability that a random male will have a

polymorphic form at a given polymorphic site that makes him incompatible as the father) is described in WO 95/12607.

[0085] If several polymorphic loci are included in the analysis, the cumulative probability of exclusion of a random male is very high. This probability can be taken into account in assessing the liability of a putative father whose polymorphic marker set matches the child's polymorphic marker set attributable to his father. This analysis can be further expanded to identify ancestral males (e.g., grandfather, great grandfather and so on). Such analysis can be useful in genealogical analysis, or in tracing the origin of ancestral man (e.g.) using samples obtained from an archeological site).

# Longer-term Family Heritage

[0086] In addition to the use in paternity testing, the polymorphisms and methods of the present invention can be used to determine relationships through a paternal lineage for multiple generations. The constancy and low mutational rate of these regions of the Y chromosome allow an individual to trace his specific ancestral lineage using the Y chromosome polymorphisms. For example, a specific residue (base) in a polymorphic site may be indicative of a population that is in or from a certain region in Europe. Assaying an individual for this polymorphism can indicate that the individual's paternal ancestors were in or descended from this particular region.

#### Correlation of Polymorphisms with Phenotypic Traits

[0087] The polymorphisms of the invention may contribute to the phenotype of an organism in different ways. Some polymorphisms occur within a protein coding sequence and contribute to phenotype by affecting protein structure. The effect may be neutral, beneficial or detrimental, or both beneficial and detrimental, depending on the circumstances. Other polymorphisms occur in noncoding regions but may exert phenotypic effects indirectly via influence on replication, transcription, and translation.

[0088] A single polymorphism may affect more than one phenotypic trait. Likewise, a single phenotypic trait may be affected by polymorphisms in different genes. Further, some polymorphisms predispose an individual to a distinct mutation that is causally related to a certain phenotype.

[0089] Phenotypic traits include diseases that have known but hitherto unmapped genetic components. Phenotypic traits also include symptoms of, or susceptibility to, multifactorial diseases of which a component is or may be genetic, such as autoimmune diseases, inflammation, cancer, diseases of the nervous system, and infection by pathogenic microorganisms. Phenotypic traits also include characteristics such as longevity, appearance (e.g., baldness, obesity), strength, speed, endurance, fertility, and susceptibility or receptivity to particular drugs or therapeutic treatments.

[0090] Correlation is performed for a population of individuals who have been tested for the presence or absence of a phenotypic trait of interest and for polymorphic markers sets. To perform such analysis, the presence or absence of a set of polymorphisms (i.e. a polymorphic set) is determined for a set of the individuals, some of whom exhibit a particular trait, and some of which exhibit lack of the trait. The alleles of each polymorphism of the set are then reviewed to determine whether the presence or absence of a particular allele is associated with the trait of interest. Correlation can be performed by standard statistical methods such as a κ-squared test and statistically significant correlations between polymorphic form(s) and phenotypic characteristics are noted.

[0091] The polymorphisms and assays of the present invention are of particular use in determining the appropriate populations for mapping complex genetic traits and/or disorders. Population choice can be crucial for the success of gene mapping for particular traits and/or disorders. Populations having a high degree of inbreeding are also useful for linkage analysis (see, e.g., Sheffield, VC et al., Trends in Genetics 4:391-6 (1998)), and the polymorphisms of the invention can be useful in determining the genetic heterogeneity of a population.

# Antibodies to Specific Polymorphisms

[0092] Polyclonal and/or monoclonal antibodies that specifically bind to variant gene products but not to corresponding prototypical gene products are also provided. Antibodies can be made by injecting mice or other animals with the variant gene product or synthetic peptide fragments thereof. Monoclonal antibodies are screened as are described, for example, in Harlow & Lane, Antibodies, A Laboratory Manual, Cold Spring Harbor Press, New York (1988); Goding, Monoclonal antibodies, Principles and Practice (2d ed.) Academic Press, New York (1986). Monoclonal antibodies are tested for specific immunoreactivity with a variant gene product and lack of immunoreactivity to the corresponding prototypical gene product. These antibodies are useful in diagnostic assays for detection of the variant form, or as an active ingredient in a pharmaceutical composition.

# Use of the Present Method to Produce a Database of Y Chromosome Polymorphisms

The polymorphisms of the invention can be used as the basis for, or combined with other such polymorphisms to provide, a general catalog of genome variation to address the large-scale sampling designs required by association studies, gene mapping, and evolutionary biology. There is widespread interest in documenting the amount and geographic distribution of genetic variation in the human species. This information is desired by the biomedical community, whose work would be greatly facilitated by a densely packed map of polymorphic markers, particularly SNPs in the NRY region, to be used to for example, identify genes associated with disease by linkage disequilibrium between sets of adjacent markers and the occurrence of disease in populations, and to characterize disease-related variation among populations.

[0094] Anthropologists and archeologists use genetic variation to reconstruct our species' history, and to understand the role of culture and geography in the global distribution of human variation. The requirements for these two perspectives seem to be converging on a need for an accessible, representative DNA bank and statistical database of human variation.

[0095] In addition, these systems have potential in both routine forensic and intelligence database applications, either in place of or in conjunction with more traditional "DNA fingerprinting" databases produced using methods such as restriction fragment length polymorphism mapping.

[0096] The invention may be embodied in computer-readable media containing an electronically, magnetically, or optically stored code representative of the markers for polymorphic regions of Table 1, and/or stored code configured to create the electronically stored representation of Table 1 and the corresponding geographic distributions for these polymorphic markers (see TABLE 3). Such databases may be produced using a variety of different data configurations and processing capabilities. Examples include, but are not limited to, logical databases, physical databases, relational databases, central configuration databases, and the like. Database structures for genomic information may be based on, for example, the database structures disclosed in U.S. Patent No. 6,229,911. In other examples, the data generated for use in the present invention may be used to create a general database such as that described in U.S. Pat. No. 4,970,672 or a relational database such as that described in U.S. Pat. No. 5,884,311. Databases containing data generated for use in the methods of the invention may also be a central configuration database for data that is shared among multiprocessor computer systems. See U.S. Pat. No. 6,014,669. Other database systems and design methodologies can be found in I. Fogg and M. Orlowska, Computers Math. Applic. (UK), (1993) 25:97-106; S. Ceri, et al., Proceedings of the IEEE (1987) 75:533-545.

#### **EXAMPLES**

[0097] The following examples are put forth so as to provide those of ordinary skill in the art with a complete disclosure and description of how to make and use the present invention, and are not intended to limit the scope of what the inventors regard as their invention nor are they intended to represent that the experiments below are all or the only experiments performed. Efforts have been made to ensure accuracy with respect to numbers used (e.g. amounts, temperature, etc.)

but some experimental errors and deviations should be accounted for. Unless indicated otherwise, parts are parts by weight, molecular weight is weight average molecular weight, temperature is in degrees Centigrade, and pressure is at or near atmospheric.

#### EXAMPLE 1

[0098] A phylogenetic tree was deduced from 167 polymorphisms from a Nonrecombining Region of the human Y chromosome (NRY)on the principle of maximum parsimony (Figure 2). Seven of the 167 polymorphisms had been detected by means other than DHPLC and were taken from the literature to demonstrate the applicability of the method of the invention to polymorphisms with less demographic specificity than those in TABLE 1. Seventy-three of the 160 polymorphisms detected by DHPLC had been reported previously. Underhill, P. A. et al Genome Res. 7:996-1005 (1997). Shen, P. et al Proc. Natl. Acad. Sci. USA 97:7354-7359 (2000). Of the remaining 87 unreported polymorphisms, 53 were discovered in a set of 53 individuals of diverse geographic origin during the screening of the unique sequences and repeat elements, other than long interspersed elements, contained in three overlapping cosmid sequences (GenBank accession nos. AC003032, AC003095, AC003097) and a few small fragments scattered throughout the NRY. Finally, 34 were detected during genotyping. In total, the marker panel comprises 91 transitions, 53 transversions, 22 small insertions or deletions, and an Alu insertion. All polymorphisms are biallelic, except a double transversion, M116, that has three alleles, A, C or T, defining quite different haplotypes. Two non-CpG associated transitions (M64 and M108) showed evidence of recurrence but generated no ambiguities when considered in the context of other markers. The primer sequences used to detect the 167 polymorphisms are given in Table 1).

#### **METHODS**

[0099] DNA samples. The ascertainment set consisted of the following 53 samples with their subsequently determined haplogroup designations: *Africa*: 3

Central African Republic Biaka II, III (1); 2 Zaire Mbuti II, III; 2 Lissongo II, III; 2 Khoisan I, III; 1 Berta VI; 1 Surma I; 1 Mali Tuareg III; 1 Mali Bozo III; Europe: 1 Sardinian VI; 2 Italian VI IX; 1 German VI; 3 Basque VI, IX (2); Asia: 3 Japanese IV, V, VII; 2 Han Chinese VII, 1 Taiwan Atayal VII, 1 Taiwan Ami, VII, 2 Cambodian VI, VII; Pakistan: 2 Hunza VI, IX; 2 Pathan VI, VII; 1 Brahui VIII; 1 Baloochi VI; 3 Sindhi III, VI, VIII; Central Asia 2 Arab IX; 1 Uzbek IX; 1 Kazak V; MidEast: 1 Druze VI; Pacific: 2 New Guinean V, VIII; 2 Bougainville Islanders VIII; 2 Australian VI, X: America: 1 Brazil Surui, 1 Brazil Karatina, 1 Columbian, 1 Mayan all X. An additional 1,009 chromosomes, representing 21 geographic regions, were genotyped by DHPLC for all markers other than those on the terminal branches of the phylogeny. The latter were genotyped only in individuals from the haplogroup to which those markers belonged. This hierarchic genotyping protocol was necessitated by the minute amounts of genomic DNA available for most samples.

[00100] PCR. The RepeatMasker2 program (<a href="http://ftp.genome.washington.edu">http://ftp.genome.washington.edu</a>) was used to identify human repeat DNA sequences. Primers were designed to amplify unique sequences and repeat elements other than LINE as confirmed by a negative female control, yielding amplicons 300-500 bp in length. All primers had a uniform annealing temperature, which allowed a single PCR protocol to be used. It comprised an initial denaturation at 95°C for 10 min to activate AmpliTaq Gold<sup>®</sup>, 14 cycles of denaturation at 94°C for 20s, primer annealing at 63-56°C using 0.5°C decrements, and extension at 72°C for 1 min, followed by 20 cycles at 94°C for 20 s, 56°C for 1 min, and 72°C for 1 min, and a final 5-min extension at 72°C. Each 50-μl PCR reaction contained 1 U of AmpliTaq Gold<sup>®</sup> polymerase, 10 mM Tris-HCl, pH 8.3, 50 mM KCl, 2.5 mM MgCl<sub>2</sub>, 0.1 mM each of the four deoxyribonucleotide triphosphates, 0.2 μM each of forward/reverse primers, and 50 ng of genomic DNA. PCR yields were determined semi-quantitatively on ethidium bromide stained agarose gels.

[00101] DHPLC analysis. Unpurified PCR products were mixed at an equimolar ratio with a reference Y chromosome and subjected to a 3-minute 95°C denaturing step followed by gradual reannealing from 95 to 65°C over 30 min.

Ten microliters of each mixture were loaded onto a DNASep<sup>TM</sup> column (Transgenomic, San Jose, CA), and the amplicons were eluted in 0.1 M triethylammonium acetate, pH 7, with a linear acetonitrile gradient at a flow rate of 0.9 ml/min<sup>2</sup>. Under appropriate temperature conditions, which were optimized by computer simulation (available at <a href="http://insertion.stanford.edu/melt.html">http://insertion.stanford.edu/melt.html</a>), mismatches were recognized by the appearance of two or more peaks in the elution profiles.

- With QIAGEN (Valencia, CA) QIAquick spin columns. Both strands were sequenced to determine the location and chemical nature of any polymorphic sites, using the amplimers as sequencing primers and ABI Dye-terminator cycle sequencing reagents (PE Biosystems, Foster City, CA). Each cycle sequencing reaction contained 6 μl of purified PCR product, 4 μl dye terminator reaction mix, and 0.8 μl of primer (5 μM). Cycle sequencing was started at 94°C for 1 min, followed by 25 cycles of 96°C for 10s, 50°C for 2s, and 60°C for 4 min. The sequencing products were purified with Centrifex<sup>TM</sup> gel filtration cartridges (Edge Biosystems, Gaithersburg, MD) and analyzed on a PE Biosystems 373A sequencer.
- [00103] Statistical analysis. The program CONTML in PHYLIP, version 3.57c, was used to construct a frequency based maximum likelihood network. The expected Luria-Delbrück/Lea-Coulson distribution of the number of mutants for each gene was fitted by maximum likelihood, treating each nucleotide of the screened sequence as analogous to a parallel, independent bacterial culture Luria, S. E. & Delbrück, *Genetics* 28:491-511 (1943); Lea, D. E. & Coulson, A. C. *Genetics* 49:264-285 (1949). The distributions under the expectation of constant population size were calculated according to Watterson, G. A. *Theor. Popul. Biol.* 7: 256-276 (1975). Mismatch distributions were calculated as described previously (Shen et al., *supra*). The NRY mutation rate per nucleotide per year (1.53 x 10<sup>-9</sup>) was calculated on the basis of 597 nucleotide substitution differences between human and chimpanzee observed over 39,931 bp of non-coding sequence (Shen et al., *supra*). The corresponding mutation rates for mtDNA (1.65 x 10<sup>-8</sup>)

and X chromosome (7.54 x 10<sup>-10</sup>) were calculated on the basis of 581 and 58 nucleotide substitution differences, respectively, between human and chimpanzee observed over 6,176 bp of coding mtDNA (mictochondrial DNA) sequence comprising the genes *ND1*, *ND2*, *COX1*, *COX2*, *ATP8*, *ATP6*, *COX3*, and *ND3*, and 7,853 bp of flanking non-coding sequence of the *DIAPH2* gene on Xq22.

[00104] Accession numbers. Most of the NRY sequence surveyed was derived from 5 cosmid sequences retrievable from Genbank using the accession numbers AC003031, AC003032, AC003094, AC003095, and AC003097. Six polymorphisms were affiliated with genomic regions for DFFRY (AC002531), one each for DBY (AC004474) and UTY1 (AC006376), 3 for SRY (NM003140), and 15 for random genomic STSs reported by Vollrath D, et al. *Science* 258:52-59 (1992).

[00105] The tree of Figure 2 is rooted with respect to non-human primate sequences. The 116 numbered compound haplotypes were constructed from 167 mutations (markers) of which 160 were discovered by DHPLC (Table 1). Seven haplotypes from the literature with less geographical heritage specificity were also analyzed in this study, including YAP (M1), DYS271 (M2), PN3 (M29), SRY 4064 (M40), TAT (M46), RPS4YC711T (M130), and SRY 2627 (M167), (the sequences for these markers are not shown in TABLE 1). Marker numbers indicated on the segments are discontinuous because of the removal of all but one polymorphism associated with tandem repeats and homopolymer tracts whose ancestral state is uncertain. Haplotypes are assorted into ten haplogroups (I - X)using principles commonly applied to haploid mtDNA phylogenies. Macaulay, V. et al. Am. J. Hum. Genet. 64: 232-249 (1999). Haplogroup I members, ancestral for M42, M94 and M139, also share the only homopolymer-associated marker M91. All haplogroup I individuals have an 8-T length variant, while 1,009 men in haplogroups II-X have 9 T's and in two cases 10 (not shown). Only one inconsistent haplogroup X individual had 8 T's (not shown). Haplogroups I and II, both of which are almost exclusively represented in Africa only, share the ancestral allele of M168. Haplogroup III is generally the most frequent one in Africa. Its frequency decreases with increasing distance from Africa, from 27% in

the Mid-East to a few percent in Northern Europe and South and Central Asia. Haplogroup IV, related to the former through M1 and M145, is found mainly in Japan.

[00106] In a recent cladistic analysis of nine diallelic NRY polymorphisms, including M1, in 1,544 individuals, it was hypothesized that haplogroup III comprises descendents of a range expansion that brought Y-chromosomes back to Africa (M. F. Hammer et al. 15:427-441 (1998)). Haplogroups V and VIII are prevalent in New Guinea and Australia, but they are also found at varying though smaller frequencies throughout Asia. Haplogroups VI and IX are found mostly in Europe and the Indus Valley. They are not observed in East Asia, where haplogroup VII dominates, suggesting that this part of the world where agriculture developed independently resisted effectively subsequent gene flow Macaulay, V. et al *supra*. The distinction between Eurasians and East Asians was also observed with mtDNA Macaulay, V. et al., *supra*., and autosomal genes (Diamond, J. *Guns, Germs, and Steel* (Norton & Co., New York, p. 99, 1999). Haplogroup X is common in the Americas, although its origin may have been in Central Asia where traces of it persist, as shown in Table 2:

TABLE 2.

Haplogroup	Exemplary Defining Mutation	Avg. no. of Mutations from Root to Individual Haplotypes	Total no. of Individuals	No. of Mutations per Haplogroup Minus Defining Mutation(s)	No. Haplotypes per Haplogroup
I	M91	6.1 ±0.95	52	20	8
И	M60	6.1±0.41	52	12	10
III	M96	10.4±0.24	218	27	21
IV	M124	10.5±0.56	9	7	4
V	M130	6.6±0.6	40	8	5
VI	M89 & absence of M9	7.4±0.25	163	25	23
VII	M175	9.5±0.35	137	18	15
VIII	M9 & Absence of M175 and M45	8.9±0.63	67	16	11
IX	M173	10.2±0.20	195	13	13
X	M74 & Absence of M173	9.2±0.1	129	6	6
Totals		8.59±0.20	1052	152	116

# EXAMPLE 2

[00107] The root of the phylogeny was placed using sequence information generated from the three great ape species. The sequential succession of mutational events is unequivocal, except for those appearing in the same tree

segment (e.g., M42, M94, M139). The phylogeny is composed of 116 haplotypes and their frequencies in 21 general populations are listed in Table 3. Forty-two haplotypes (36.2%) are represented by just one individual. Several haplotypes, however, display higher frequencies and/or geographic associations that reveal patterns of population affinities apparent from a maximum likelihood analysis (Figure 3) performed on the haplotype frequencies reported in Table 3. To facilitate presentation, the 116 haplotypes were grouped into 10 haplogroups as defined either by the presence or absence of mutations occupying strategic positions in the phylogeny. Haplogroups VI, VIII, and X, although polyphyletic, are distinguished by the criteria in Table 2.

[00108] Three mutually reinforcing mutations, M42, M94 and M139 (2 transversions and a 1-bp deletion) unequivocally distinguish haplogroup I which is represented today by a minority of Africans, mainly Sudanese, Ethiopians, and Khoisans (Table 2). All non-African, except a single Sardinian, and the majority of African males sampled, carry only the derived alleles at the three sites. This implies that modern extant human Y-chromosomes trace ancestry to Africa and that the descendents of the derived lineage left Africa and eventually replaced archaic human Y-chromosomes in Eurasia.

[00109] An important property of a phylogeny is the randomness of number of mutations per segment of the tree. Forty-one of the total 166 segments carry no mutation, while 98, 16, 8, 2, and 1 segment have 1, 2, 3, 4, and 8 mutations, respectively. The mean number of mutations per segment is 1.024 with a variance of 0.945. Applying the G-test for goodness of fit and Williams' correction to the observed G, the data do not fit a Poisson distribution (Gadj=34.98, df=3, P~10<sup>-7</sup>). This is due to an excess of segments with one mutation, as expected in an exponentially growing population. Similar results were obtained recently for the separate analysis of 4 Y-chromosome genes. Further support that the human population has undergone a major expansion comes from the consistently negative values of Tajima's D (Lea, DE & Coulson, AC Genetics 49: 264-285 (1949)) for not only the Y-chromosome, but also for mitochondrial DNA, X-

chromosomal and autosomal genes. Interestingly, NRY shows evidence of significantly reduced variability to the other genetic systems (Shen et al., supra), confirming a similar comparison of a smaller number of polymorphisms on previously reported NRY sequences with eight X-linked (Hudson, R. et al, Genetics 116:153-159 (1987); Nachman, M. W. Mol. Biol. Evol. 15: 1744-1750 (1998) and 16 autosomal human genes. Possible explanations include positive selection on NRY Jaruzelska, J et al., D. Mol. Biol. Evol. 16:1633-1640 (1999) and a difference between male and female effective population sizes Wyckoff, G. J et al., Nature 403:304-309 (2000). Assuming expansion, the age of the most recent common ancestor ( $T_{MRCA}$ ) was previously estimated at 59,000 years with a 95% probability interval of 40,000-140,000 years (Thomson, R. et al. supra).

[00110] This value is similar to an estimate of 46,000 to 91,000 years based on 8 Y chromosome microsatellites (Pritchard, J. K et al, *Mol. Biol. Evol.* 16:1791-1798 (1999) and, therefore, is considerably less than estimates of >100,000 years obtained previously (Hammer et al, *supra*). Of course, this assumes that selection or population structure have not had a major effect on NRY diversity, an assumption that may be wrong in light of our findings of significantly reduced variability on NRY. As the average number of mutations of all segments departing from the root is 8.60 (Table 3), and with a  $T_{MRCA}$  value of 59,000 years, the average time for adding a new mutation to the tree is 6,900 year. This puts the age of M168 that marks the expansion of anatomically modern humans out of Africa at approx. 44,000 years, in agreement with a previous estimate of 47,000 years with 95% probability intervals of 35,000 to 89,000 years using the program GENETREE (Thomson, R. et al. *Proc. Natl. Acad. Sci.* USA 97:7360-7365 (2000).

TABLE 3.

D-d-t-t																																											
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Sardinia	l	1																																	1	•		4	•				
Basque Mid-east	1																								2						1				1			2	,		1	1	
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Pakistan + India	1													2																				2			1						
Hunza	1																																										
Japan	ľ																																										
China	1																																										
Taiwan	l																																										
Cambo + Laos	l																																										
New Guinea	ł																																										
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Taiwan							1																				2					1				4	1		18		1		
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New Guinea		•		•		-	7								2	5	4	1													•										1	23	
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[00111] This concurs with recent archeological and mtDNA data, and is also consistent, though at a compressed time scale, with the weak Garden of Eden hypothesis. Under this hypothesis, a small subgroup of behaviorally modern humans left Africa and separated into several fairly isolated groups represented today by the major haplogroups III-X. Those groups remained small throughout the last glaciation before they underwent roughly simultaneous expansions in size as suggested by the star-like genealogy shown in Figure 1. In conclusion, the new levels of biallelic variation revealed here suggest a recent ancestry of the paternal lineages of our species from Africa and testify to the informativeness of the Y chromosome in deciphering the evolution of humankind.—

[00112] The gene frequencies of New Guineans and Australian aborigines were grouped together because of the small sample size of the latter. Values at nodes indicate number of 1,000 bootstrap trees presenting cluster distal of node. Sudanese and Ethiopians are distinct from the other Africans and appear to be more associated with samples from the Mediterranean basin. This may reflect either repeated genetic contact between Arabia and East Africa during the last 5,000 to 6,000 years or a Middle Eastern origin with subsequent acquisition of Negroid genes on the way southwest with agricultural expansion. Native Americans are located between Eurasians and East Asian indicating common ancestry with both. This network is consistent with the first two principal components capturing 18% of the variation present in the 116 haplotypes.

#### EXAMPLE 3

[00113] A phylogenetic tree was deduced from NRY polymorphisms on the principle of maximum parsimony (Figure 3). Figure 3 shows the phylogenetic tree deduced from 304 polymorphisms including those presented in Examples 1 and 2 as well as other novel markers.

[00114] The contemporary global frequency distribution of the 10 Groups based on >1000 globally diverse samples genotyped using a hierarchical top down approach is illustrated in Figure 3. 171 haplotypes are identified in Fig.3 as well as their relationship with 309. However 4 markers are recurrent but define

distinctive haplotypes when considered in the context of the other markers. The 4 markers are M64.1 (M64.2),M108.1 (M108.2), M116.1 (M116.2) and 12f2.1 (12f2.2). For example M64.1 occurs on haplotype #80 in Group V and M64.2 on ht#159 in Group IX.

[00115] The relationship of the haplotypes to the ten haplogroups is also shown in Fig. 3. Each haplotype can be related to a specific geographical region within the haplotype group, allowing for very specific geographic association and ethnic identity of male individuals. Fig. 3 also shows which specific markers are important branching points for distinguishing between haplotype groups and also sub-haplotype groups such as haplotypes 10-13 of group II. This composite collection of 315 NRY variants (polymorphic markers) provides improved resolution of extant patri-lineages.

#### **EXAMPLE 4**

[00116] The methods of the invention can be utilized in the area of forensics to determine the ethnic affiliation of an individual.

[00117] The method involves, obtaining a nucleic acid sample from the individual and processing the sample sufficiently to conduct PCR amplification on the sample. The method exploits the hierarchical property of the Y chromosome gene tree that reveals the unequivocal sequential accumulation of DNA variation during the lineal life spans of these haplotypic molecules. Since Y chromosome haplotypes display a strong correlation with geography, such data provides insights into the affinity and diversification of populations. The sample is analyzed at polymorphic sites defining key internal nodes within the phylogeny. At least 11 primers sets, with each primer set recognizing at least one polymorphic region on the Y chromosome from a different haplotype group (Group I through Group X) are required to begin localizing a sample within the phylogeny. Additional haplotype resolution can be obtained by typing a subset of related markers. Each PCR reaction carried out on the sample, may include one or more primer sets/reaction vessel.

[00118] The PCR amplified products are analyzed by DHPLC (or any other suitable PCR product detection technique, such as DNA chips, direct sequencing, Taqman and the like) genotyping technology to define the haplotype which is then compared to a data base detailing the geographic association of the haplotype. The data base utilizes the markers identified in TABLE 1 and various combinations thereof which enables the identification of an individual to a particular haplotype group (Group 1 through Group X) as well as haplotype which are indicated in FIG.2 and FIG.4.

[00119] In certain instances, primer sets to the following markers are utilized to identify which haplotype group an individual originates from; Markers- M91, M60, M96, M174, (M216 or M316), M89, M9, M175, M45, M173. These markers identify the following haplotype groups; Group I = M91, Group II = M60, Group III = M96, Group IV = M174, Group V = M316, Group VI = M89 without M9, Group VII = M9 without M175 or M45, Group VIII = M9, Group IX = M173 and Group X is represented by marker M74 without M173. This approach can be expanded to increase criteria for inclusion/exclusion decisions.

[00120] TABLE 4 shows a two stage scheme of 30 markers, the haplotype groups they help define as well as geographical region associated with the haplotype group and the polymorphic markers which provides considerable power in facilitating localization any Y chromosome in the phylogeny. In cases where more than one marker is listed in TABLE 4, any one marker in the subset will provide comparable information.

TABLE 4			
Markers analyzed	Assoc. Geographical	Markers analyzed	Assoc.
Analysis #1	region	Analysis # 2	Geographical
			region
M42, M94, M251, or	Global	M215, M243, or	Africa, Med
M299 (Groups II-X)		M293 (Group III)	
M246	Africa	M2, M180 or	Sub Saharan Africa
(Group I)		M291 (Group III)	

M181 or M249 (Group II)	Africa		Sub Saharan Africa					
		M191 (Group III)	Suo Sanaran Amea					
M168 or M294	Global	M35	A.C.: - N I					
	Global		Africa, Med,					
(Groups III-X)		(Group III)	S. Europe					
M96	Africa, Med.	M217	E. Asia, India,					
(Group III)		(Group V)	N. America,					
M174	Asia, Japan	M201	Med., S. Europe					
(Group IV)		(Group VI)						
M216 or M316	Asia, America,	M172	Med., S. Europe					
(Group V)	Polynesia, Melansia	(Group VI)	*					
M89, M213 or M235	Global	M267	Med., S. Europe					
(Groups VI-X)		(Group VI)						
M9	Global	M170 or M258	Europe					
(Groups VII-X)		(Group VI)						
M175 or M214	E. Asian	M52 or M69	India					
(Group VII)		(Group VI)						
M45 or M74	Global	M122	E. Asia					
(Groups IX-X)		(Group VII)						
M173 or M207	Europe, W. Asia	M119	E. Asia					
(Group IX)		(Group VII)						
M269	W. Europe	M268	E. Asia					
(Group IX)		(Group VII)						
M242	C. Asia, India,	M17 or M198	E. Europe, W. Asia					
(Group X)	Americas	(Group IX)						
M304	Med.	M3	N.& S America					
(Group VI)		(Group X)						

[00121] This example demonstrates that by using about 10% of the markers, one can localize any sample to a "neighborhood" or sub-haplotype group in the tree. These markers are useful in identifying a male for which no ethnic origin is

known. If it was known that the individual to be typed was for example, from Peking, then the assemblage of a more "Asian" group of markers would be more useful than those in TABLE 4.

- [00122] The methods of the invention allow for the ability of Y markers to define (on a general geographic or population level) male ethnic affiliation.
- [00123] While the present invention has been described with reference to the specific embodiments thereof, it should be understood by those skilled in the art that various changes may be made and equivalents may be substituted without departing from the true spirit and scope of the invention. In addition, many modifications may be made to adapt a particular situation, material, composition of matter, process, process step or steps, to the objective, spirit and scope of the present invention. All such modifications are intended to be within the scope of the claims appended hereto.

#### TABLE 1

M2 = DYS271 (209 bp) A to G at position 168

aggcactggtcagaatgaagTGAATGGCACACAGGACAAGTCCAGACCCAGGAAGGTCCAGTAACATGGGAGAAGAACGGAAGGAGTTCTAAAATTCAGGGCTCCCTTGGGCTCCCCTGTTTAAAAATGTAGGTTTTATTATTATTATTTCATTGTTAACAAAAGTCCRTGAGATCTGTGGAGGATAAAGGggggagctgtattttccatt (SEQ ID NO:1)

For: 5'-3' = aggcactggtcagaatgaag (SEQ ID NO: 2)

Rev 5'-3' = aatggaaaatacagctcccc (SEQ ID NO: 3)

M3 = DYS199 (241 bp) C to T at position 181

taatcagtctcctcccagcaAGTGATATGCAACTGAGATTCCTTATGACACATCTGAACA CTAGTGGATTTGCTTTGTAGTAGGAACAAGGTACATTCGCGGGATAAATGTG GCCAAGTTTTATCTGCTGCCAGGGCTTTCAAATAGGTTGACCTGACAATGGGT CACCTCTGGGACTGAYAATTAGGAAGAGCTGGTACCTAAAATGAAAGATGCc cttaaatttcagattcacaatttt (SEQ ID NO: 4)

For: 5'-3' = taatcagtctcctcccagca (SEQ ID NO: 5)

Rev 5'-3' = aaaattgtgaatctgaaatttaagg (SEQ ID NO: 6)

M4 = DYS234 (273 bp) A to G at position 88

 $tcctaggttatgattacagagcgAGGATTATTATAATATTGGAATAAAGAATAATTGCTACA\\ AACTAATGATTAATGATATTCATAT<math>m{R}$ TAATCATATCTAAGATCTATATCTAGTATATCTTATTTTATTTTATTTTATTGTACTGGAACAGCTTGTGCCCTTGG

TCTCTTGCCTCGGCACCTGGGTGGCTTGCCATCCACAGAAGTGTTTTAACAGC AAAAATTACTGTGAATTTCTGCCCAAAAccttgtcatgtttacaagacgt (SEQ ID NO: 7)

For: 5'-3' = tcctaggttatgattacagagcg (SEQ ID NO: 8)

Rev 5'-3' = acgtcttgtaaacatgacaagg (SEQ ID NO: 9)

M5 = DYS214a (322 bp) C to T at position 73

gggtttatactgacctgccaatgttAAAAGGGACCTAAATTCACTTTGGGGAAGTGGCCAGA AAGGAAGAAGYAGAAGGAGAAGAGAGAAACCTCCAGTTGTGGGGGTT GAGCCTCCAGGATAAGAAAGAAAGAAATCTCCAGTAGGGGGGATTGAGCCT AACACAAACCTTTGGTAATAGACAAGGCAAGACATTTCCAATAGGGGAGATT GAGTGTCACCTCAAAACTATTAAGATGGGAAATACCCCAGGTAAGATAGAGG GTAAAAAAGGATAAAGCTAGCAGCAATAACATTCcccctgaaagttcccaataa (SEQ ID NO: 10)

For: 5'-3' = gggtttatactgacctgccaatgtt (SEQ ID NO: 11)

Rev 5'-3' = ttattgggaactttcagggg (SEQ ID NO: 12)

**DYS214 complete.** (656 bp) This fragment was converted into two STSs, a & b, containing M4 and M16 respectively. The two new STSs (a & b) omit an extra internal 68 bp region within the complete STS.

For 3'-5' = gggtttatactgacctgccaatgtt (SEQ ID NO: 14)

Rev 5'-3' = ccgtgtgttgctgggctgtc (SEQ ID NO: 15)

M6 = DYS198 (218 bp) **T** to **C** at position 37

CactaceacatttctggttggCTTGTAGTTCTTTCTYGGAAAAATATTATTCTAATTTCCTT ATAGTATTAGCCATCAAAGTAGGGGAAGCAGATCAAATCTACCATAAGACCA AGTCATAGGAAGAAGATCAAATTAAGATGCTAGGCAAAAGTCTCAGCACATA TGGATTATGAGAAGCACATTCACACATCCAAActcaaagaatggactcagcg (SEQ ID NO: 16)

For: 5'-3' = cactaceacatttctggttgg (SEQ ID NO: 17)

Rev 5'-3' = cgctgagtccattctttgag (SEQ ID NO: 18)

M7 = DYS253 (300 bp). C to G at position 236

For: 5'-3' = actgtgagcgagctgaaaat (SEQ ID NO: 20)

Rev 5'-3' = gcagccttgtgaaccaatta (SEQ ID NO: 21)

# M8 = DYS263 (267 bp). G to T at position 137

CccaccacttcagtatgaaTTTTGGGATCTGTTACCTATTTTTTGATATAAAATCAACTG CAAGTTTAGTGCCTCAGTATCACAAACACTGTATTTGCTCATATGTCTGTGAA TCAATAACTTGGACTGGGTTCA**K**TTGGGCAGTTCTTCTATTGGTCTTGCCTGG GGTCTTTAATGCAGCTTCCATTTTCTGGCAGCTTGATGAGACTGGATGGTCTA AGGTACATTCATGAACACACTCTGTTTGgtggacttgtctgtcagcct (SEQ ID NO: 22)

For: 5'-3' = cccacccacttcagtatgaa (SEQ ID NO: 23)

Rev 5'-3' = aggctgacagacaagtccac (SEQ ID NO: 24)

# M9 (340 bp) G10.35a C to G substitution at position 68

GcagcatataaaactttcaggACCCTGAAATACAGAACTGCAAAGAAACGGCCTAAGAT
GGTTGAATSCTCTTTATTTTTCTTTAATTTAGACATGTTCAAACGTTCAATGTC
TTACATACTTAGTTATGTAAGTAAGGTAGCGCTTACTTCATTATGCATTTCAA
TACTCAAAAAAAATTCCTTTGTGAAATGTTGAAATATTTTTCTAATCTGTTTC
ACGAGCTTCAAAAAATGAGGAAAAAAGATTCAGTTTACATTTCAGCAAAAATGC
CTCTTTTTAATCGGATTTATGTTTACTTAACATTTACAGTACATTTACgcttgagcaa
agttaggtttt (SEQ ID NO: 25)

For: 5'-3' = geageatataaaaettteagg (SEQ ID NO: 26)

Rev 5'-3' = aaaacctaactttgctcaagc (SEQ ID NO: 27)

# M10 = G10.10 (343bp). T to C at position 156

For 5'-3 = gcattgctataagttacctgc (SEQ ID NO: 29)

Rev 5'-3' = taataaaaattgggtcaccc (SEQ ID NO: 30)

# M11 = G10.37 (222 p) A to G at position 44.

TetetetgtetgteteteceteeCTCTCTCTTGTATTCTAAC**R**GAAAGGTTTAGAACTTGCA TAATTGGGAAAGAAGCTGTTGCCTGAACTTACTGGGGGATTCAGCATTGTCA TTTTGGACATGTCACTTATCCTCAGTATTTGCTTCCCCCAGGAGAGAGCTGTA ATAAAAAAGCATTGCAATTTAATACATAAgetcagtaagttcttgtttatgete (SEQ ID NO: 31) For: 5'-3' = tetetetgtetgtetetecetee (SEQ ID NO: 32)

Rev 5'-3' = gagcataaacaagaacttactgagc (SEQ ID NO: 33)

M12=DYS260a (309 bp) G to T at position 286

ATTATTTGTAGA**K**ATGggggtcactatgttgctcag (SEQ ID NO: 34) For: 5'-3' = actaaaacaccattagaaacaaagg (SEQ ID NO: 35)

Rev 5'-3' = ctgagcaacatagtgacccc (SEQ ID NO: 36)

M13 = G10.06 (233 bp) G to C at position 157

TcctaacctggtggtctttcATTGTTTTACAAAGGTGATTTAGTTTTGGGAAGGACTATTC TCCTTTAAACTATAGACTAAATTTTTCTCAAAGTTAGGTTAGTTTATGCCCAG GAATGAACAAGGGCAGTAGGTAGGTTAAGGGCAAGACGGTTASATCAGTTCT CTGTTACTGTTATAATTTTCTCATTGTTATATTTTTTGCAAATGTGgttggataaaatca tggctca (SEQ ID NO: 37)

For: 5'-3' = tcctaacetggtggtctttc (SEQ ID NO: 38)

Rev 5'-3' = tgagccatgattttatccaac (SEQ ID NO: 39)

M14 = G10.07 (287 bp) **T to C** at position 180

For: 5'-3' = agacggttagatcagttctctg (SEQ ID NO: 41)

Rev 5'-3' = tagataaaagcacattgacacc (SEO ID NO: 42)

M15 = G10.16 (295 bp = ancestral state); derived allele = 9 bp insertion (304 bp) after position 109; Note that there are also two T to G changes immediately before the 9 bp insertion.

AcaaatcctgaacaatcgcCATCACCTATTTGGTGGACGCATAGGCCTGGTCTCTGATCT GGTCGCATGTCCAGAGGGTCTGCTAACCCACTGCACCTAGGGAGACA**TTGTA CAGAGA**CATTGTACCACCTTTTCTCTACTcttcccagactcaacacatttGATTGTATATGC GCATGAGGTAGAAATATAAGATGAAGCAGGGACAGAGTCAACAAGCCAGAA CTAGATGCTTCTACCTGGACAGAAGACCTAGAATTCTTTTTTGGATCCTAAAT TCACCAggaaattttaaccacatgca (SEQ ID NO: 43)

For: 5'-3' = acaaatcctgaacaatcgc (SEQ ID NO: 44)

Rev 5'-3' = tgcatgtggttaaaatttcc (SEQ ID NO: 45)

M15 polymorphic region in more detail

mutant sequence = GACA TT GTACAGAGA CA (SEQ ID NO: 46)

ancestral sequence = GACA GG \*\*\*\*\*\* \* CA (SEQ ID NO: 47)

#### M16 = DYS214b (266 bp) C to A

TgttatgtcatttgaacccagGAACCAATCTTCGAAC**M**CTCAGTTTTCTGGCCAAAGTTGGAGTCAAATGAGGATTGGATTTGTCAGCTTTTAATAGAACATATGATGACAAAACCTTCATCTCCCAGGAGGAGATAAATTATGCCCTATGTTGGTGGCAAGGACCTGTCCTCCTTTACCCTCTAAAAACTGGAGGAGAAAGTCAAAGACTAACTCCTCTGAAAAAGATAAAGTCCCTATTCCTAgacagcccagcaacacacgg (SEQ ID NO: 48)

For: 5'-3' = tgttatgtcatttgaacccag (SEQ ID NO: 49)

Rev 5'-3' = ccgtgtgttgctgggctgt (SEQ ID NO: 50)

M17 = G10.47a (333 bp) -1bp deletion (4G's to 3G's) at position 68

CtggtcataacactggaaatcAGATTCTGTCTACTCACCAGAGTTTGTGGTTGCTGGTTGT TACGGGGTTTTTTTAAGTGAATTTTTGGGGTTTGTTAAGTGGCCAAACTATTTT TGTGAAGACTGTTGTATGTGGGTTTCAGATGTCTCTACATCAGTTTGTGGTCA GCTAGTGAGTTAAATTTTATGAAAAGCCTGGAGAAACAAGAATAGCAGTAAA AACTTCCAGTCTTTGTAGATTGGGTGTCTTCAGTGCTTAGCTGGGCAATTTAA AACTTACCTTAAGTAGTACAGTTGGCCCTTTGTGTCTTGTgagtttcacatttgtaggttca (SEQ ID NO: 51)

For: 5'-3' = ctggtcataacactggaaatc (SEQ ID NO: 52)

Rev 5'-3' = tgaacctacaaatgtgaaact (SEQ ID NO: 53)

M18 = G10.47b (333 bp = ancestral size) +2 bp (extra AA) insertion after position 62 CtggtcataacactggaaatcAGATTCTGTCTACTCACCAGAGTTTGTGGTTGCTGGTTGT TAAACGGGGTTTTTTTAAGTGAATTTTGGGGTTTGTTAAGTGGCCAAACTATT TTTGTGAAGACTGTTGTATGTGGGTTTCAGATGTCTCTACATCAGTTTGTGGT CAGCTAGTGAGTTAAATTTTATGAAAAGCCTGGAGAAACAAGAATAGCAGTA AAAACTTCCAGTCTTTGTAGATTGGGTGTCTTCAGTGCTTAGCTGGGCAATTT AAAACTTACCTTAAGTAGTACAGTTGGCCCTTTGTGTCTGTgagtttcacatttgtaggttc a (SEQ ID NO: 54)

For: 5'-3' = ctggtcataacactggaaatc (SEQ ID NO: 55)

Rev 5'-3' = tgaacctacaaatgtgaaactc (SEQ ID NO: 56)

M19 = G10.47c (333 bp) **T** to **A** at position at 131

ctggtcataacactggaaatcAGATTCTGTCTACTCACCAGAGTTTGTGGTTGTTACGGGGTTTTTTAAGTGAATTTTGGGGTTTGTTAAGTGACCAAACTATTTTTGTGAAGACTGTTGTAWGTGGGTTTCAGATGTCTCTACATCAGTTTGTGGTCAGCTAGTGAGTTAAATTTTATGAAAAGCCTGGAGAAACAAGAATAGCAGTAAAACTTCCAGTCTTTGTAGATTGGGTGTCTTCAGTGCTTAGCTGGGCAATTTAAAACTTACCTTAAGTAGTACAGTTGGCCCTTTGTGTCTGTgagtttcacatttgtaggttca (SEQ ID NO: 57)

For: 5'-3' = ctggtcataacactggaaatc (SEQ ID NO: 58)

Rev 5'-3' = tgaacctacaaatgtgaaactc (SEQ ID NO: 59)

M20 = G10.48. (413 bp) A to G at position 118

For: 5'-3' = gattgggtgtcttcagtgct (SEQ ID NO: 61) Rev 5'-3' = cacacaacaaggcaccat (SEQ ID NO: 62)

M21 = G10.43 (415 bp) A to T at position 357

For: 5'-3' = cttttatttctgactacaggg (SEQ ID NO: 64)

Rev 5'-3' = aacagcagatttgagcagg (SEQ ID NO: 65)

M22 = DYS273 (327 bp) A to G at position 129.

AgaagggtctgaaagcaggtTCGTGATTTCACCCTTTACAGTTTAATACAAGGGATTTTA CATACAGACATATAAGCTGATAGTCCTGGTTTCCCTATTTGTTTTAAGGTGCC ATTCCTGGTGGCTCTRCCTCCTCCCCAGTGCCCATATGGGCCCTTAGTCTG CTGTAGGCATGCTCAGGCAAGCCCTTGAGCAAATTCCCTTAATCTGCACGAA ACATGGGCTGGAGATTCAGTGGGACCCTTTCTTTAGTGTCTGCCTAATGCAAG CTGGCTAACTCCTTTCAAAAGTTTTGTCTTGCTGATgaagcctccaggtagtaggc (SEQ ID NO: 66)

For: 5'-3' = agaagggtctgaaagcaggt (SEQ ID NO: 67)

Rev 5'-3' = gcctactacctggaggctt (SEQ ID NO: 68)

M23 = G10.57a (327 bp) A to G at position 159

For: 5'-3' = tctctaacttctgtgagccac (SEQ ID NO: 70)

Rev 5'-3' = ggaaaaactaaactctaaatctct (SEO ID NO: 71)

M24 (tetranucleotide TAAA motif) = SRY 8299c. Internal primer regions for SRY4064 which contain M40 and M41.

For: 5'-3' = acagcacattagctggtatgac (SEQ ID NO: 73)

Rev 5'-3' = tctctttatggcaagacttacg (SEQ ID NO: 74)

M25 = B9.008b. (340 bp) **G** to **C** substitution. Position 121

AaagcgagaattcaatccagGATGACAGAATGCGTTCACCTTTAAAGGGATTAAAAGA AGTATAATACAGTCTGTATTATTAGATCACCCAGAGACACACAAAACAAGAA CCGTGAATTSAATTAGTGGTATACTAATAGAGTGGTTTTACCTGAAATATTTA CACATCAATCCTACTGAATTCTTACAACAAATGATTTAGATTAGCTATTGTAT TCACCAGTTGAAAGAACAGAAAATATTGAGGGAGATAACTTGTGTCAGTGCA ACTTAATCAGATTTAGGACACAAAAGCAACTACATAATGAAAAAGAGAgctggt gacttaacttgctaaaa (SEQ ID NO: 75)

For: 5'-3' = aaagcgagagattcaatccag (SEQ ID NO: 76)

Rev 5'-3' = ttttagcaagttaagtcaccagc (SEQ ID NO: 77)

M26 = B9.005 (321 bp) G to A at position 68

CcagtggtaaagttttattacaatttTTTTAAACCAAGATTCAATTTTTTTCTGAATTAGAATT ATC**R**CAGAGAACACTGAATGGCCTATGAAATTCAATTTTTGCTGCAGATTTC GTCATGTTTCTTAATGAACATATAACTAACTTAATCACAAGATAAATTCTT GCCTATGTGCAAAAACTTAGTGCTGCATCCTTGTGTATGGTTTTAAAAAGTGT CAAAACTGGCCCCTCATGTCAAATACAGCCCCAATTAGGGGAGGCAACCTAA GAAAGGTGTACAACTGTCCTGACATTggattgcctgcttactgtgaa (SEQ ID NO: 78)

For: 5'-3' = ccagtggtaaagttttattacaattt (SEQ ID NO: 79)

Rev 5'-3' = ttcacagtaagcaggcaatcc (SEQ ID NO: 80)

M27 = G10.65. (526 bp). C to G at position 398.

GTGTTGAGAACCAGACAATACGAAATAGAAGACAAATCATGAGCTTACAGA ACCTGAAACTTTTTACACTGGGCAGtgtggtagacagaacagcagtg (SEQ ID NO: 81)

For: 5'-3' = cggaagtcaaagttatagttactgg (SEQ ID NO: 82)

Rev 5'-3' = cactgctgttctgtctaccaca (SEQ ID NO: 83)

**M28**= G10.33n (332 bp). **T to G** at position 277.

GcttacttgggacacaggctAGTTCTCTCCTGAAGCTATTGAGCAGTATGTTGAGGTG CGCTACGCCAGTTGAGGTGAAGCTGTTACACAGTATGAAAGCCGGGCTTTGT AGCTGCAGCTGCGCATTGCACCCCCAGCTACGCAGTCTCCTTTCCTTCTCAGT CACAGGACCGGATGGCAAGTGGCCGCAGCCAGTCGGTGAGACCGACTGAGC TCTGGGGCTTCAGTTCTTGACGCTACCTACATGGCTACATCTCCAGCCAAGGA TGAGAGGKGATGCCAGAGGACCTCGATCTAAATTGGGCAccattatcgtatgacaacttct ct (SEQ ID NO: 84)

For: 5'-3' = gcttacttgggacacaggct (SEQ ID NO: 85)

Rev 5'-3' = agagaagttgtcatacgataatgg (SEQ ID NO: 86)

M30 = G10.66 a (486 bp) G to A at position 132.

For: 5'-3' = gaaccagacaatacgaaatagaag (SEQ ID NO: 88)

Rev 5'-3' = tttagcggcttatctcattacc (SEQ ID NO: 89)

M31 = G10.66 b (486 bp) G to C at position 71.

For: 5'-3' = gaaccagacaatacgaaatagaag (SEQ ID NO: 91)

Rev 5'-3' = tttagcggcttatctcattacc (SEQ ID NO: 92)

M32 = G10.68a (370 bp) T to C at position 166.

TtgaaaaaatacagtggaacAAAGATCCTCTGTATCTCTGCTCCTAAGATAGCAGAGACA GCatactggcttctgttcaattttcCTTTGATTACACAACTTCATTGGCTACGGTGTTTAATAT GACCGTCATAGGCTGAGACAAGATCTGTTCAGTTTATCTCAYAAGTTACTAG TTAAATCTCAGACATATTATACTTTTGTAACTGAGTGACTCCCATTGTAAGGA TAACTACTTCAATGTGCGTATAAATGAGTCAGTTGTCTCTCTTGGGGGCTTCA ACAAATAAGCAAAGATAACCTCATTGTGGAGAGCACTTCACATTTGTTTTTAG GGTTACATAGTCTActctgtatccttaaacacttg (SEQ ID NO: 93)

For: 5'-3' = ttgaaaaaatacagtggaac (SEQ ID NO: 94)

NewF 5'-3' = atactggcttctgttcaattttc (SEQ ID NO: 95)

Rev 5'-3' = caagtgtttaaggatacaga (SEQ ID NO: 96)

M33 = G10.68b (370 bp) A to C at position 180.

TtgaaaaaatacagtggaacAAAGATCCTCTGTATCTCTGCTCCTAAGATAGCAGAGACA GCatactggcttctgttcaattttcCTTTGATTACACAACTTCATTGGCTACGGTGTTTAATAT GACCGTCATAGGCTGAGACAAGATCTGTTCAGTTTATCTCATAAGTTACTAGT TAMATCTCAGACATATTATACTTTTGTAACTGAGTGACTCCCATTGTAAGGA TAACTACTTCAATGTGCGTATAAATGAGTCAGTTGTCTCTCTTGGGGGGCTTCA ACAAATAAGCAAAGATAACCTCATTGTGGAGAGCACTTCACATTTGTTTTTAG GGTTACATAGTCTActctgtatccttaaacacttg (SEQ ID NO: 97)

For: 5'-3' = ttgaaaaaatacagtggaac (SEQ ID NO: 98)

New F 5'-3' = atactggcttctgttcaattttc (SEQ ID NO: 99)

Rev 5'-3' = caagtgtttaaggatacaga (SEQ ID NO: 100)

M34 = G10.69 (372 bp) G to T at position 131

CactteacatttgtttttaggGTTACATAGTCTACTCTGTATCCTTAAACACTTGAAGATCT GTTATAACTACATCTGAGATAGTCACAGTGTTTTCTCATGTTAATGCCTG GCTTCCACCCAGGAGKCACATGTGGTGTGTCTGCAAATAAAGTGTTTATGAT TATTGGGGTCCCCCAAGCTGGACCTGTATCCATGTTCAAGTGGCCACAGGGTT ACTTGCTTTAGCATGGCTCCTTGGCTGGCTGTTAAGTGAATAATTAAACTGAG TCTTTTTTGCAGGAGCTAACTGAGACCAATCAATCAGTCAATTTTCCCTTTCT GTGTGTAACACAAGCTGGATGTCCctggaatgactaaataatgact (SEQ ID NO: 101)

For 3'-5' = cacttcacatttgtttttagg (SEQ ID NO: 102)

Rev 5'-3' = agtcattatttagtcattccag (SEQ ID NO: 103)

M35 = G10.72a (514 bp) G to C at position 168

For: 5'-3' = taagcctaaagagcagtcagag (SEQ ID NO: 105)

# Rev 5'-3' = cagagggagcaatgaggaca (SEQ ID NO: 106)

M36 = G10.82a (436 bp) T to G at position 74

For: 5'-3' = agatcatcccaaaacaatcataa (SEQ ID NO: 108)

Rev 5'-3' = aaggctgaaatcaatccaatctg (SEQ ID NO: 109)

M37 = G10.STS 84 (422 bp) C to T at position 203. This STS also contains M61 at position 101 which is defined in G10.83.

For: 5'-3' = cagattggattgatttcagcctt (SEO ID NO: 111)

Rev 5'-3' = agcatacaaaaaaaaaaaaaactgc (SEO ID NO: 112)

M38 = G10.73a (337 bp) T to G at position 146

For: 5'-3' = cagtttttagagaataatgtcct (SEQ ID NO: 114)

Rev 5'-3' = ttaaagaaaagaaaagcagatg (SEQ ID NO: 115)

M39 = G10.73a (337 bp) - 1 bp (-C) deletion at position 236

# GCCCCCATTATATGATGATACCTTGTAATGATTTAATTTTAGcatctgcttttcttttaa (SEQ ID NO: 116)

For: 5'-3' = cagtttttagagaataatgtcct (SEQ ID NO: 117)

Rev 5'-3' = ttaaagaaaagaaaagcagatg (SEQ ID NO: 118)

M41 = SRY 4064b (218 bp) G to T at position 117. Site is located within SRY 8299 509 bp STS.

GtataataggctgggtgctgTGGGTCACACCTGTAATCCCAGCCCTTCGAGAGGTCAAGG CAAGCGGATCACAGGGTGGAAGAGATTGAGACCATCCTGGCCAACATGGTG AAACTKGGTCTCTACTAAAAATACAAAAAATTAGCTGGGCGTGGTGACATGT GCCTGTAATCCCAGTTACTCGGGAGGCTGAGGCAGaagaatcatttgaactcatg (SEQ ID NO: 119)

For: 5'-3' = gtataataggctgggtgctg (SEQ ID NO: 120)

Rev 5'-3' = catgagttcaaatgattctt (SEQ ID NO: 121)

# M42 = B9.008a (340 bp) A to T substitution at position 297

For: 5'-3' = aaagcgagagattcaatccag (SEQ ID NO: 123)

Rev 5'-3' = ttttagcaagttaagtcaccagc (SEQ ID NO: 124)

#### M43 = DYS260b (309 bp) A to G at position 77

For: 5'-3' = actaaaacaccattagaaacaaagg (SEO ID NO: 126)

Rev 5'-3' = ctgagcaacatagtgacccc (SEQ ID NO: 127)

#### M44 = G10.87 (389 bp) G to C at position 263

For: 5'-3' = ctggcaccttctgatattttgag (SEQ ID NO: 129)

# Rev 5'-3' = tgtgatttctatgtgtttgaggac (SEQ ID NO: 130)

#### M45= B9. 12(352 bp) G to A substitution at position 109

GctggcaagacacttctgagCATCGGGGTGTGGACTTTACGAACCAACCTTTTAACAGTA ACTCTAGGAGAGAGGATATCAAAAATTGGCAGTGAAAAATTATAGATA**R**GC AAAAAGCTCCTTCTGAGGTCCAGGCCAGGAGATAGTAGGATTTAAGAAACAA ACAAACAAAAAACAACCACAAATGACCTTTGGTGCCACTGTCACAACTGTTGC TCATCAGAGTAGGAGAGTTGTAGCAAAGGCATTAAAGAAGGACAAGCT GAAGAGCCTGAATCCTTGTGTTGTAAGCTATTTTGGTTTCCTTTCAAGAAAGG GCTGTGGTCTGTggaaggtgtcaggaacatatt (SEQ ID NO: 131)

For: 5'-3' = gctggcaagacacttctgag (SEQ ID NO: 132)

Rev 5'-3' = aatatgttcctgacaccttcc (SEQ ID NO: 133)

#### M47 = G10.82b (436 bp) G to A at position 395

AgatcatcccaaaacaatcataaCTTGTTTAAATTGTTCATAGCAAAAGTTACATATTATA AAGAGTTATGAGTGTCTTAGGCAGTGAATAGCAAAAGTTACCTTTTATAGT TGTCCTTCACTAGCAGGAAGCCTTATTCCCTGCCCTTTTACATATCTTAACTTA GAATGTTACTGTCTAAATAGTGGTTAGGCAAGAGTAGTTCTTAAACGTGCAG TAATTATCTTGCACTACATTTAAGGGCTAAATAGCTAGTAGTGGTGCTTGATA ATTGAAGAAATTTGTACAGCTGGAGGAAGTACCTGCTAAATTTTCAAAAGTT ACCTGAATTTAATAGGTAAATCTGTTTTTAATTAGAGCTATATCATTTTACTCT GAATGTCTTAACATARAAATTTACATATAACATARAAATTTACAGAGttgatttgatttcagcctt\_(SEQ\_ID\_NO: 134)

For 5'-3'= agatcatcccaaaacaatcataa (SEQ ID NO: 135)

Rev 5'-3' = aaggetgaaatcaatccaatctg (SEQ ID NO: 136)

#### M48 = G10.79n (240 bp). A to G at position 160

For: 5'-3' = aaacaatatgtatgctaattttgct (SEQ ID NO: 138)

Rev 5'-3' = tcaatgtaaatgttagtataaggatg (SEQ ID NO: 139)

#### M49 = B9.15 new a (354 bp) **T** to **C** at position 229

For 5'-3'= cggcaacagtgaggacagt (SEQ ID NO: 141)

Rev 5'-3' = tgcttcaggagatagaggctc (SEQ ID NO: 142)

# M50 = B9.15new b (354 bp) **T** to **C** at position 175

CggcaacagtgaggacagtAGCTCCAGGTCTGGGCGGAAGGTGGTGCGGTGAAAGGTGCAGGACAGACTGGGTTAGAGGCCACTCTTGGTCTTATCCTCCATGGCCACAACAGAGGTGACAAATACATGGGTCACTCAGTTATGTTTAGCCAACAGCCTACCCAAACCACACCYGTCTTACCAGAGCCCTTTCCTGGAGCCATGTTCTCAGGACTGGTCACACTGTCTCCATTCTCCAGCAGCCCTTGGACCTATCGGAAAAAAAGAATGGGTAACAATAATTGAGCTGATGAACCAGGTCCTATCTTCCTCCACAACTCCAAAACTTGGgagcctctatctcctgaagca (SEO ID NO: 143)

For: 5'-3' = cggcaacagtgaggacagt (SEQ ID NO: 144)

Rev 5'-3' = tgcttcaggagatagaggctc (SEQ ID NO: 145)

#### M51 = B9.16 (339 bp) G to A at position 33

For: 5'-3' = gagcetetateteetgaage (SEQ ID NO: 147)

Rev 5'-3' = tgactgctctgttgcgaca (SEQ ID NO: 148)

# M52 = G10.88 (534 bp) A to C at position 477

For: 5'-3' = actgtagcatggtcatctagggtg (SEQ ID NO: 150)

Rev 5'-3' = gacgaagcaaacatttcaagagag (SEQ ID NO: 151)

# M53 in tree (tetranucleotide TAAA motif) = SRY 8299d. Internal primer regions for SRY4064 which contain M40 and M41.

CTACCCTATATTCTAAAATTCAAAAGTAATGGTTTTTGTTATGAAATCTcgtaagt cttgccataaagaga (SEQ ID NO: 152)

For: 5'-3' = acagcacattagctggtatgac (SEQ ID NO: 153)

Rev 5'-3' = tctctttatggcaagacttacg (SEQ ID NO: 154)

#### M54 = B9.17 (360 bp) G to A at position 164

For: 5'-3' = cctcctctggtctgggttt (SEQ ID NO: 156)

Rev 5'-3' = tgtgtcaggactggttccat (SEQ ID NO: 157)

#### M55 = B9.28 (382 bp) T to C at position 228

For: 5'-3' = cgtaggcgtttgacagcag (SEQ ID NO: 159)

Rev 5'-3' = cetttettegtaateeteee (SEQ ID NO: 160)

#### M56 = B9.29 (399 bp) A to T at position 39

CcagaaactgaagtacaaatgcAATGGGAGGATTACGAWGAAAGGAGGGCTAAGTGAT GATAAGTATGGTCAGAATAAATAAATTTATTCTAGACAAGAAATGAGAGTTCA TTATGTCAGAAGCAAAATAGTACTACAGGATGACAACTTCTGAGATTTACTCT TTGGTTCCAACTGCCTACAAGACAAAGAAAAACTGAAGAGGCCAGGAAGTTAA ATGCATGAGGAAAACTTGAGGCAGATTAAAATGGAAATGCAGGGCATGTTAT TTGGGTATCATGGGTTCAATCTGGAAAAGCCTTATTTCTCCTGAACCACAGTA GGGAAAGGAGTTATCCAGAAAAGTGAAATTTATCTAAAATTTTAAGTTTCC ATGTTTTaaagagggcagcaatgaga (SEQ ID NO: 161)

For: 5'-3' = ccagaaactgaagtacaaatgc (SEQ ID NO: 162)

Rev 5'-3' = teteattgetgeetetettt (SEQ ID NO: 163)

M57 = G10.85n (326 bp ancestral); +1 bp insertion (327 bp = Derived). Extra A inserted at position 133

ACTATACGATTTGAAACAAAATTCTTTTTTTGGAGACTATGGAAACATTCTCA ACAGGGAAACCCTACTAGACTTTGTAAAgcaaataatggaaaagatacagaac (SEQ ID NO: 164)

For: 5'-3' = attgggaggaagtggtttctg (SEQ ID NO: 165)

Rev 5'-3' = gttctgtatcttttccattatttgc (SEQ ID NO: 166)

# M58 = G10.57b (327 bp) G to A at position 224

For: 5'-3' = tetetaaettetgtgagecae (SEQ ID NO: 168)

Rev 5'-3' = ggaaaaactaaactctaaatctct (SEQ ID NO: 169)

#### M59 = B9.15 new c (354 bp) A to C at position 279

For: 5'-3' = cggcaacagtgaggacagt (SEQ ID NO: 171)

Rev 5'-3' = tgcttcaggagatagaggctc (SEQ ID NO: 172)

# M60 = B9.34 (388 bp ancestral); +1 bp insertion (389 bp = DERIVED). Extra T inserted after position 242

GeactggcgttcatcatctGGGAGCAGCTCAAAAGCCTCTCGCTCAGCCTCCGTGACGCC CTGGGGGTGTTCAACCCACATATACTGTAAAGACTAGGAGTAGGGTTGTGGA CACCCCACCTCAGCCAACACTGAGCCCTGATGTGGACTCAACCTTGTAAGGA AAGCTGTAGAGAAAATTGGAAGAAAAAATATAAACACATACAGACTCTGTCTT TACATTTCAAAATGCATGACTTAAAGTATCAGGCACACAGTGGTTACTCAAT GTTGGTCTGTGTCTCTGTAACGTAATATATGTGACTAAATCCCTAAGCTCTGC TCTTGACCACCCACCTTCTCCAAAAAGGGCCTTTCGTAGACGTCGCTcctcctgaacca taatgaacat (SEO ID NO: 173)

For: 5'-3' = geactggcgttcatcatct (SEQ ID NO: 174)

Rev 5'-3' = atgttcattatggttcaggagg (SEQ ID NO: 175)

#### M61 = G10. 83 new a (190 bp) C to T at position 98.

For: 5'-3' = attggattgatttcagcette (SEQ ID NO: 177) Rev 5'-3' = attttatttctgtgttccttgc (SEQ ID NO: 178)

#### M62=DYS260c (309 bp) T to C at position 60

For: 5'-3' = actaaaacaccattagaaacaaagg (SEQ ID NO: 180)

Rev 5'-3' = ctgagcaacatagtgacccc (SEQ ID NO: 181)

# M63 = B9.22 (308 bp) G to A at position 43

For: 5'-3' = ctcttcccttggttcctattc (SEQ ID NO: 183)

Rev 5'-3' = ttaggcagaagcatccacc (SEQ ID NO: 184)

# M64 = B9.t23 (325 bp) A to G at position 279 RECURRENT

TatagaccetgactactcaagagaaAAGTCCAATCCAAAGAAAAAATACAAAAGAAAACA
AAATCACATCAGGCCACAAACCAGTTTAAGGGCCCTCACCACATGGTTGGCT
CCAGACTGAAACATTTCATAGGGGTAAATAATGCGTTCGTAATGTGATCGTA
GCAGGGAGCCAATGTTTTTGCCTGGTGGGTAGTGGAGACGCTGGGCAACTCG
AGCCCACCGACGATCCTTGCAGATGGCTTCATAGCCACCTTCCTCAATCACAA
TCTGAAAGTRTAAGAAACAATATGGATGAACTGTGAacagactggaaagggctacc
(SEQ ID NO: 185)

For: 5'-3' = tatagaccctgactactcaagagaa (SEQ ID NO: 186)

Rev 5'-3' = ggtagccctttccagtctgt (SEQ ID NO: 187)

#### M65 = B9.t26 (436 bp) A to T at position 152

TtctgatgccagcttgttcgGGTCAGAAAAGTTAAATGAGAAATTTGGTGCTAAGGGTTTCTGGTCATGAGTGTAAATAACGCCTCGCCAAGTGGTAAACTGCCCCAACGTTCAAACCAAAGGCTACCCATTCCCAAATTTTGTTTCAAAGWCTTACCGCGGGTGGGCGGATTTTGCAGATGCCAGACTTCTCTGCTATGGGCCTTATTTTCGCAATGTAGCCAAGCGGGTCTTGGAATTCAGCCCAGCTAGGCTCAAAAACCGGGCACTCCGGTGGCGGCAGGAACTCGTCACACCCCGGTTCCATGTCGGGCCTTAATGCTAAGCTGTAAAAATAAGAATCACATTGTCTTTAATGACGCGCTGGTTCCTCCTACTAAAAAGGCCTATGAAAATTTCATTTTCTTGAGAATTTCaaggttactttaatcccgtagc(SEO ID NO: 188)

For: 5'-3' = ttctgatgccagcttgttcg (SEQ ID NO: 189)

Rev 5'-3' = gctacgggattaaagtaaccttg (SEQ ID NO: 190)

### M66 = B9.41 (415 bp) A to C at position 135

For: 5'-3' = ctgtgtaacaccatcaagtgc (SEQ ID NO: 192)

Rev 5'-3' = acatettetggagacatacttec (SEQ ID NO: 193)

#### M67 old = B9.36 new a (409 bp) A to T at position 377

CcatattctttatactttctacctgcAGGCCCACTGCATGCTCACTCACCCAGTCAGCAGTACA AAAGTTGACAGCTTCAGCAAAATTGTAGCCTTGGTTAAAACCACTGTGGTAA GCACGAGGAAAAGTGATGACAAACTCCCCTGCACACTGGTTTGTGCGGACAA CCTAAAAAAGGAGAAAAAAAGCAGAAAAGAGGTGTGGGTCAGAACTAATGGGCC AGATGTGAACTCAAAGATGTCTCTAGATGCTGTAACAGATGTAGGAAGAGTG GAAAGGCTCTATCTTCAAGTACGTGTCCTAAAAAGAAAAATGAGATTGTGAAT TTAAAAGTGGTATTCATAGAAAAAGTACTCAAAAATATGTGTAATTCAAAAAAAC

AWATATAGAGGGtccacgaacaagtgaaaagac (SEQ ID NO: 194)

For: 5'-3' = ccatattetttataetttetaeetge (SEQ ID NO: 195)

Rev 5'-3' = gtcttttcacttgttcgtggac (SEQ ID NO: 196)

#### M67 revised B9.36new a (386 bp) STS A to T at position 327

ccagtcagcagtacaaaagttgACAGCTTCAGCAAAATTGTAGCCTTGGTTAAAAACCACTG TGGTAAGCACGAGGAAAAGTGATGACAAACTCCCCTGCACACTGGTTTGTGC GGACAACCTAAAAAGGAGAAAAAAGCAGAAAGAGGTGTGGGTCAGAACTAA TGGGCCAGATGTGAACTCAAAGATGTCTCTAGATGCTGTAACAGATGTAGGA AGAGTGGAAAGGCTCTATCTTCAAGTACGTGTCCTAAAAGAAAATGAGATTG TGAATTTAAAAGTGGTATTCATAGAAAAGTACTCAAAATATGTGTAATTCAA

AAAACAWATATAGAGGGTCCACGAACAAGTGAAAAGACTCTttgcttctataatcaa agaaatgc (SEQ ID NO: 197)

newFor 5'-3' = ccagtcagcagtacaaaagttg (SEQ ID NO: 198)

newRev 5'-3' = gcatttctttgattatagaagcaa (SEQ ID NO: 199)

#### M68 old = B9.36 new b (409 bp) A to G at position 268

CcatattctttatactttctacctgcAGGCCCACTGCATGCTCACTCACCCAGTCAGCAGTACA
AAAGTTGACAGCTTCAGCAAAATTGTAGCCTTGGTTAAAACCACTGTGGTAA
GCACGAGGAAAAGTGATGACAAACTCCCCTGCACACTGGTTTGTGCGGACAA
CCTAAAAAGGAGAAAAAAGCAGAAAGAGGTGTGGGTCAGAACTAATGGGCC
AGATGTGAACTCAAAGATGTCTCTAGATGCTGTAACAGATGTAGGAAGRGTG
GAAAGGCTCTATCTTCAAGTACGTGTCCTAAAAGAAAAATGAGATTGTGAAT
TTAAAAGTGGTATTCATAGAAAAAGTACTCAAAATATGTGTAATTCAAAAAAAC
AAATATAGAGGGgtccacgaacaagtgaaaagac (SEQ ID NO: 200)

For: 5'-3' = ccatattetttatactttetacetge (SEQ ID NO: 201) Rev 5'-3' = gtetttteacttgttegtggae (SEQ ID NO: 202)

M68 revised B9.36new b (386 bp) STS A to G at position 219

newFor 5'-3' = ccagtcagcagtacaaaagttg (SEQ ID NO: 204) newRev 5'-3' = gcatttctttgattatagaagcaa (SEQ ID NO: 205)

M69 = B9.62a (257 bp) T to C at position 222

GgttatcatagcccactatactttgGACTCATGTCTCCATGAGAACTAAGACTACCACAACA GAATCCCTATAGTCCAGCCCTCAGATCACATACATGTACAGGCATGTTGAAG TAGTCGGACTTGAAGGAATCAGCCATTTCACCAAAACTCTGCAAACTGTACT CCTGGGTAGCCTGTTCAAATCCAAAAGCTTCAGGAGGCTGTTTACACTCCTGA

AAYAAAATATTTCagcaagacaaagggaataaagat (SEQ ID NO: 206)

For: 5'-3' = ggttatcatagcccactatactttg (SEQ ID NO: 207)

Rev 5'-3' = atctttattccctttgtcttgct (SEQ ID NO: 208)

M70 = B9.62b (257 bp) A to C at position 45

GgttatcatagcccactatactttgGACTCATGTCTCCATGAGAMCTAAGACTACCACAACA GAATCCCTATAGTCCAGCCCTCAGATCACATACATGTACAGGCATGTTGAAG TAGTCGGACTTGAAGGAATCAGCCATTTCACCAAAACTCTGCAAACTGTACT CCTGGGTAGCCTGTTCAAATCCAAAAGCTTCAGGAGGCTGTTTACACTCCTGA AATAAAATATATTTCagcaagacaaagggaataaagat (SEQ ID NO: 209)

For: 5'-3' = ggttatcatagcccactatactttg (SEQ ID NO: 210)

Rev 5'-3' = atctttattccctttgtcttgct (SEQ ID NO: 211)

M71 = B9.63b (328 bp) C to T at position 197

TtgaattatagtcccttgcctcTGGTTCAGTCAAGTCTCTATCATTCTAGAGTTAGTGTGTT CAATCGTTCTTGTATAGTAGCTCACTGATAGCTTAATCAAAACCTAACACAAA TATTAACTTATAAAAGGGCAGAAACTACCTTCCCAAAACCCAGAAGGGGAGA TTACAGAAAATCACCAACCAAAAATAAAGYATCTGTGACAGACAGATCTTAC CGCCAAGATACATTTTGGGCACCTCCAGATGCCTCTGGGGATTTCAGGAAGG GGTGGTAACAAGCAGAAGATGTGGTAATTGTCATCAcagccatcacagaaaagaagc (SEQ ID NO: 212)

For: 5'-3' = ttgaattatagtccettgcete (SEQ ID NO: 213)

Rev 5'-3' = gcttcttttctgtgatggctg (SEQ ID NO: 214)

M72 = B9.63a (328) A to G at position 157

For: 5'-3' = ttgaattatagtcccttgcctc (SEQ ID NO: 216)

Rev 5'-3' = gcttcttttctgtgatggctg (SEQ ID NO: 217)

# M73 = B9.47a (361 bp ancestral & 359 bp derived) -2bp deletion,

(-GT) at position 260

cagaataataggagaatttttggtCAAATAAAAGGCCATATTATATTTCTTTTGATAAAAGT ATCATGTGTTCAGTATGTTTTATTATTTTGAAATAATTAACATGACAGGAATAT ATTTGAAAAAAAATTCCAAAAAAAAGCTAAATATACAAACTAAGAAAAATTATAT GATTATACTTATCTGCAGTATTGTAAAAACAATAGTTCCAAAAAACTTCTGAATT ACAAGTTTAATACATACAACTTCAACTTCAACTACATTGTAGACGTTC AGAGGAATCACAAAGGACCTCAACATGCTAGATAAGAAAATGTATTTTTAA ATGTTTTGGCTCAgctgcttagaaaataaggaaaat (SEQ ID NO: 218)

For: 5'-3' = cagaataataggagaatttttggt (SEQ ID NO: 219)

Rev 5'-3' = attttccttattttctaagcagc (SEQ ID NO: 220)

M74 = B9.50a (385 bp) G to A at position 195.

For: 5'-3' = atgctataataactaggtgttgaag (SEQ ID NO: 222)

Rev 5'-3' = aattcagcttttaccacttctgaa (SEQ ID NO: 223)

M75 = B9.51 (355 bp) G to A at position 296

GctaacaggagaaataaattacagacTGTAAAAGTTGATGACCAAGAATTTTTCAGAAGTGG TAAAAAGCTGAATTCTCAAGTTTGAGAATTCCTATCTATTCCCAGAAATATTAA GTAAAAAGTCACATTCCACACATCAAGAAAACTTGCAAGACACTAAAAGAG ATATTATAGCAGTCAAATAGAAAAAGCAAAATAGACTACTACAAATTAATGT AAGATTCAGAATTGACTTGTCAAAAGCCAAAACAGATTTCTAATGTACTGTG AAAAGACAATTATCAAACCACATCC**R**TATATATACAGAGAAATACCTTTATA AGAATAAAAATtcacaaatgcctctgttcaata (SEQ ID NO: 224)

For: 5'-3' = gctaacaggagaaataaattacagac (SEQ ID NO: 225)

Rev 5'-3' = tattgaacagaggcatttgtga (SEQ ID NO: 226)

M76 = G10.100a (493 bp) T to G at position 339

For: 5'-3' = tagaagtagcagattgggagagg (SEQ ID NO: 228) Rev 5'-3' = cctgataaaatgaaaaaatggtc (SEQ ID NO: 229)

M77 = G10.105 (371 bp) C to T at position 129

CttttcttccttagetgttccTTTCCTGTGGTTTTAAAAAAAGTGACCAGAAACTAGGTCTCT ATTTTCATTGCTTGCTGCATATTCTTTTAACCTGCTTTTATCTTTTACAGAGTT GAGGGGCTTTYTAAAAACCTAGACAATGTCAAGATTCTTAGCTGCGTTTTCT GTCTAAAAGTGTAGATGTCTAGTTATTCCTCATGTAAAACACAACATTTCAAC CCTGAGTACTATAAACTTTATTATGCTTCTAGGTTACTTTTTCTCTTTAAGCAA TTATTCCTACATTCCTAAGTGTTCACCAGTGGAACAGATAAGAGATAGAAGT AGTTAGAAATTGAGATAATTGggttgacctgtcattgttgc (SEQ ID NO: 230)

For: 5'-3' = cttttcttcccttagctgttcc (SEQ ID NO: 231) Rev 5'-3' = gcaacaatgacaggtcaacc (SEQ ID NO: 232)

M78 = B9.60a (301 bp) C to T at position 197

CttcaggcattattttttttggtTCTCCACTACAGGAGAAATGTAAATGTGATGAGTCAGAAT TTAGGATGGCTGTATGGGTTTCTTTGACTAATACAAGAAATCACTTTGTAATG AATGAAATCAGTGGTTTCTGCATTACTCCGTATGTTCGACATGAACACAAATT GATACACTTAACAAAGATACTTCTTTCYGCCCTTCCAAATATTTCAAAATAAG CTGGTCATAGTACTTGCTTTTCATAAAAAAGATGGTAAGCTTCCAATATTTAGA TTTaaggaaaggtgaaggaacactat (SEQ ID NO: 233)

For: 5'-3' = cttcaggcattattttttttggt (SEQ ID NO: 234) Rev 5'-3' = atagtgttccttcacctttcctt (SEQ ID NO: 235)

M79 = B9.42 Homopolymer in tree (425 bp = majority men). A's. 8 A's to 9 A's (426 bp derived). Extra "A" inserted after position 212.

For: 5'-3' = agccagttggatgacacgtt (SEQ ID NO: 237)

# Rev 5'-3' = cctcttcgtttcctgacattt (SEQ ID NO: 238)

**M80** = G10.107. **Homopolymer in tree** (290 bp = most men). 9 T's to 10 T's (291 bp derived). Extra "T" inserted after position 55.

For: 5'-3' = actttetettetttttagggtgace (SEQ ID NO: 240)

Rev 5'-3' = acacacacacacacacacatcttatgg (SEQ ID NO: 241)

M81 = B9.58a (422bp) C to T at position 147.

ActtattttatagtttcaatccctcaGTAATTTTAACTTACTTCTATTTTAAGAACTATAACCA AACTATCTGTAAGACTTTTAAGCACTATCATACTCAGCTACACACTCTCTTAAC AAAAGAGGTAAATTTTGTCCTTTTTTGAAYGTCATAGAGTATACTCACACAA ACCAAGAAGAAACAATCTACTACATACCTACGCTATATGGTATATAACTATT GCTCCTAGGCTACAAATTAGTGCGACACTATTGTACTGAATATTATAGGCCAT GTAACACAAATGGTTTAAGTATCTGTGCCTCTAAACACAGAAAAGATATAGTG AAAGTACAGTATTGCTCCTTTATTAAACTCAAAATGTTATGCAGCATATGACC GACTATAAAATAGCGCTTATCcagatacagacatctccatgaa (SEQ ID NO: 242)

For: 5'-3' = acttaatttatagttteaatecetea (SEQ ID NO: 243)

Rev 5'-3' = ttcatggagatgtctgtatctgg (SEQ ID NO: 244)

For: 5'-3' = ctgtactcctgggtagcctgt (SEQ ID NO: 246)

Rev 5'-3' = aagaacgattgaacacactaactc (SEQ ID NO: 247)

M83 = B9. Alu01 (503 bp) C to T at position 120

GggaaaggagttatccagaaaAGTGAAATTTATTCTAAAATTTTAAGTTTCCATGTTTTA AAGAGAGGCAGCAATGAGAAAAAAGGTTAAGAACAAGTAGGAAATACTGAA ATAATGGGYCAGGCACGGTGGCTCATGCTTGTAATCCCAGCACTTTGGGAGG CCAAGGCAGGCAGATCACAAGGTGAGGAGATTGAAACCATCCTGGCTAACAT GGTGAAACCCCATCTCTACTAAAAATACAAAAAAATTAGCCAGGTGTGGTGG CACACACCTGTAGACCCAGCTACTTGGGAGGCTGAGGCAGGATAATGGCCTG AACCCGGGAGGTGGAGCTTGCAATGAGCTGAGATCGTGCACTCCAG

For: 5'-3' = gggaaaggagttatccagaaa (SEQ ID NO: 249)

Rev 5'-3' = aatgacccatgcttacttagc (SEQ ID NO: 250)

**M84** = B9.72 **Homopolymer in tree**(439 bp = most men). 9 T's to 8 T's (438 bp derived). One deleted "T" at position 400.

For: 5'-3' = ccctctccaactgagttcaag (SEQ ID NO: 252)

Rev 5'-3' = gcaatatacgtttctgcttcca (SEQ ID NO: 253)

M85 = B9.67a (568 bp) C to A at position 437

For: 5'-3' = aacagaattatcaggaaaaggttt (SEQ ID NO: 255)

Rev 5'-3' = gcaatatacgtttctgcttcca (SEQ ID NO: 256)

M86 = B9.t25a (324 bp) T to G at position 85

TcccattatttgctatatttgctACATACATCTAAGGTCATATCAAAGAAAGAAAACACCAG TCCAAGTGGTTAACACACACAGC**K**TATATAACTTGCTTCTGTCATAGATCAAG TACTTCTGAGTAAGCTATTTTTTTGCGGTTAAATGTAATAAAAGCTTGTGTAT GCCTAAACTATATTTAATAACAGCAGAACGTAGAAATATTTGAATCTTATATT TTTGTCCCTACAGCAGTCAGATGTTTAGAACCCCGTGGAATGTGGCGATCTGA TAC**T**AATATTCTGATGCCAGCTTGTTCgggtcagaaaagttaaatgagaaa (SEQ ID NO: 257)

For: 5'-3' = teccattatttgetatatttget (SEQ ID NO: 258)

Rev 5'-3' = tttctcatttaacttttctgaccc (SEQ ID NO: 259)

M87 = B9.t25b (324 bp) T to C at position 277

TCCAAGTGGTTAACACACACAGCTTATATAACTTGCTTCTGTCATAGATCAAGT ACTTCTGAGTAACACACACAGCTTATATATAACTTGCTTCTGTCATAGATCAAGT ACTTCTGAGTAAGCTATTTTTTTTGCGGTTAAATGTAATAAAAGCTTGTGTATG CCTAAACTATATTTAATAACAGCAGAACGTAGAAATATTTGAATCTTATTT TTGTCCCTACAGCAGTCAGATGTTTAGAACCCCGTGGAATGTGGCGATCTGAT ACYAATATTCTGATGCCAGCTTGTTCgggtcagaaaagttaaatgagaaa (SEQ ID NO: 260)

For: 5'-3' = teccattatttgetatatttget (SEQ ID NO: 261)

Rev 5'-3' = tttctcatttaacttttctgaccc (SEQ ID NO: 262)

M88 = B9.80 (314 bp) A to G at position 166

AttctagggtcaggcaactaggGAATACTGCTGTAGCCTAGAGCCTGCCAAAATTATTCA AACTAGCCAATCCCATACTTCTTATCCTGCTCTTGTCTTGCCTTTCCCTTGGTAA ACCCAATATAGGCTATGGCCTAGGTGCTTTTCTTATTCCTGCTTCTTCTGCRT ATCCAAGATAGGTTTTCCTCTCTAGCACTGTGTAGCATATAGTGACTACCTCT CTAAGGCCTGTGATAATAATAAAACTTTGCTTTCCTGAGTCTCTGTGGTCACAC CTACTGACCATCACATggaagaccatagaatagaacaaaca (SEQ ID NO: 263)

For: 5'-3' = attctagggtcaggcaactagg (SEQ ID NO: 264)

Rev 5'-3' = tgtttgttctattctatggtcttcc (SEQ ID NO: 265)

**M89**= B9.94 (527 bp) **C** to **T** at position 347

For: 5'-3' = agaagcagattgatgtcccact (SEQ ID NO: 267)

Rev 5'-3' = tccagttaggagatccctca (SEQ ID NO: 268)

M90 = B9.96 (331 bp) C to G at position 170

TgatgtttcttcagtctttgaggTTGCTGTCTTTTTGGATTTTTGAAAAAAATCCTATTTAATAA CTTAGTGGGTTGGTTTGTAGCAACAGTGAATTCAATCAACTGGCTTTATTCT AGAATATTTTAAAGATATTTTATCTCAGGATTTCTGGATGGTGTTCTGTAACT STAGGGACTGGGAATGAGCTTTGGCTTTGTTCCTTTACACCCTGAGGTTAGAA ATCTGCTGCACTGGAGGGACCAAGATGCTCTCAGAGAAAATGGTCACAACACT CTAATGATTGGTAGTAGCCAATGTGCTTCATATGCGggtggtagcaggattcatctt (SEQ ID NO: 269)

For: 5'-3' = tgatgtttcttcagtctttgagg (SEQ ID NO: 270)

Rev 5'-3' = aagatgaatcctgctaccacc (SEQ ID NO: 271)

**M91** = B9.87a **Homopolymer.**(495 bp, most men = 9 T's). Either one T deleted or inserted at position 368 (i.e. 8 T's or 10 T's)

For: 5'-3' = gagettggactttaggacgg (SEQ ID NO: 273)

Rev 5'-3' = aaactttaaggcacttctggc (SEQ ID NO: 274)

# M92 = B9.G2 (470 bp) T to C at position 340

TtgaatttcccagaattttgcAATCTGATCCAAATAGTTCAATTTCACTCTAGTTTGGGCCT GGGAAAGAGGGGCCTTATAAGATTGGCATACTCCTTAACCTGACTTCATCG AGTATGCAGTAAATGAACAAGTATTATTCTATGCTATCTACACTTCTCCACCA ACGTGCCGGAGCCCCAGCTTCACTGTCTTATCTCACCAGCGGGGTCCACAAA AAGCTCAAATAAGCTGAGTCTTAATCTATAAAGAGCTAAGAATGTGCCGTC TTAGGATCAACATCATGTCTAAATTTAAGGAATTATTCTTGGACTTAAAGGTG GCTTGACCAAAAAATAYGTAGGCTCCAACAGTATTTAGACTCAATATCATCAA GACACTCATTTAGAATGTACTGATATATAAATTCAAAGAATTAAAATTTTTC TAGTTCATGTAAAAGAGCTggacacaaaaccagtttctgaa (SEQ ID NO: 275)

For: 5'-3' = ttgaatttcccagaattttgc (SEQ ID NO: 276)

Rev 5'-3' = ttcagaaactggttttgtgtcc (SEQ ID NO: 277)

#### M93 = B9.93 (504 bp) C to T at position 459

For: 5'-3' = aacaaaacaaaacaaaaatactgaa (SEQ ID NO: 279)

Rev 5'-3' = ggttcacttgaagatagtttaggtta (SEQ ID NO: 280)

#### M94 = B9.122 (405 bp) C to A at position 227

CacatggagaacagagaaatgcAGTGCAGGGCAAGGGCCCACCCAGAAGCAACACAGTCAATGGAGCCTCCTTCACCCAGGAAACTGCAAACTGAATGCATGATCCTAGGATCCTCCCATGGATCTTTGCAACTTTCAGGTCAGGAGATCCAGTCAGGGACC

CATTCCACTAGGGCCTTCAGTTAGAAACACAGAGCTCATGGAGTCTTATCAG AGTAGCTGTT**M**AGGCATGCATAGGGACCCAGGAGCTTTATACACCCTGACCG TAAAGTCCCCAGCAAATATGACTGAAATTCAAGCAAGGTGGAACACTAACCT TTGCACATACACTTGGGAAGGGAGTGGAAATCAAGATGCCAAGCAGCATTGG TCTGTGAACCccactttcacaacatttcacaag (SEQ ID NO: 281)

For: 5'-3' = cacatggagaacagagaaatgc (SEQ ID NO: 282)

Rev 5'-3' = cttgtgaaatgttgtgaaagtgg (SEQ ID NO: 283)

#### M95 = B9.123 (480 bp) C to T at position 172

For: 5'-3' = gagtggaaatcaagatgccaag (SEQ ID NO: 285)

Rev 5'-3' = gggctttctgtaacctggaga (SEQ ID NO: 256)

M96 = G3.05a (440 bp) G to C at position 70. Internal lower case denotes location of alternative reverse primer region to amplify site a only, as 212 bp STS.

GttgccctctcacagagcacTTTAAAGTGAGCTGTGATGTGTAACTTGGAAAACAGGTCT CTCATAATASGATAAAACACTCAGGTATAATATTAAAAACCTATGGCAAAAT ATATGGTCCTTTACAAAGCAACAAAGTGGGTGGGTGAATCTCTTCATTCTTGG CTGGCCATCAGTTCCTGTTACTGTACaggagtgggaaaacagtagccCTGGGAAATGGGT TAAAACTGAGTAGGCATCTCCTGTGTCCAATAAGAACTCAATATTTTTGTCTG CTATATCAAGGGTTACTTGAGGCTCCTCTGTGGAGATGGTAAGTTGTCCAGTG GGAGATATAGAGAATGTTAGGCCTTATAGGTTCTCTACTTTTTTTGGCCATTAT GAGTCTGAATGTCTCAAACTCCCTTTTTATCCTGGTgcaatccttccagtgacctt (SEQ ID NO: 287)

For: 5'-3' = gttgccctctcacagagcac (SEQ ID NO: 288)

Rev 5'-3' = aaggtcactggaaggattgc (SEQ ID NO: 289)

# M97 = G3.05b (440 bp) T to G at position 355

gttgccctctcacagagcacTTTAAAGTGAGCTGTGATGTGTAACTTGGAAAACAGGTCT CTCATAATAGGATAAAACACTCAGGTATAATATTAAAAACCTATGGCAAAAT ATATGGTCCTTTACAAAGCAACAAAGTGGGTGGGTGAATCTCTTCATTCTTGG CTGGCCATCAGTTCCTGTTACTGTACAGGAGTGGGAAAACAGTAGCCCTGGG AAATGGGTTAAAACTGAGTAGGCATCTCCTGTGTCCAATAAGAACTCAATAT TTTTGTCTGCTATATCAAGGGTTACTTGAGGCTCCTCTGTGGAGATGGTAAGT TGTCCAGTGGGAGATATAGAGAATGTTAGGCCKTATAGGTTCTCTACTTTTTT GGCCATTATGAGTCTGAATGTCTCAAACTCCCTTTTTATCCTGGTgcaatccttccagt gacett (SEQ ID NO: 290) For: 5'-3' = gttgccctctcacagagcac (SEQ ID NO: 291) Rev 5'-3' = aaggtcactggaaggattgc (SEQ ID NO: 292)

For: 5'-3' = gaatggggtgttacatggaga (SEQ ID NO: 294) Rev 5'-3' = cctattatgagagacctgttttcc (SEQ ID NO: 295)

**M99** = G3.04b (395 bp nominal) **1 bp deletion** (3A's to 2A's) at position interval 96-98, STS alos has polymorphic (GTTTT) motif

For: 5'-3' = gaatggggtgttacatggaga (SEQ ID NO: 297) Rev 5'-3' = cctattatgagagacctgttttcc (SEQ ID NO: 298)

M100 = G3.04c (395 bp nominal) in tree (penta microsatellite) (GTTTT)5; (GTTTT)6 = most men); (GTTTT)7; (GTTTT)8 alleles detected

For: 5'-3' = gaatggggtgttacatggaga (SEQ ID NO: 300) Rev 5'-3' = cctattatgagagacctgttttcc (SEQ ID NO: 301)

M101 = A8.05a original (460 bp) C to T at position 154

For: 5'-3' = tcacagcagcttcagcaaa (SEQ ID NO: 303)

new R 5'-3' = ataaaaattagactctgtgttactagc (SEQ ID NO: 304) (used with F primer, just amplifies (369 bp) the first 2 sites including homopolymer T region

Rev 5'-3' = cctttttggatcatggttctt (SEQ ID NO: 305)

# M102= B9.101 (480 bp) G to C at position 301

AaactgggacacttgtaatgaatAATTACTTTGTTTGTAAATCACAATAGAGATTCTCCATA TCAAAGCTGTAACTATTCTATAGTATTTAGGCAAATAAGATAGCTACAA ATTTAAGTACTGTAATAATAGATGCCTGACAATATGTGCTATAGGTAAAATCTT TGAAATTTAATAAATGAAGTATAGATTGAATACAAGTAATATGTAATAATAC ATTATAAATTAAACATTTAGAATAATACAATTTATACAAAAAATAAAATTA AGAtaaaattcacatagtgcaatggtgASTAAGATGTGAAAAGACAATAAGAATAAACAGC ATTAAAATTATTGATAGAGATTTGTAAAACCCCTAGAGATTAAGGAAAACAAA CATAGGAATAAATTAGAAAACTAGAGACAATAATAATTCTGTAAAATTATAG GCTACCAAAACCAGAATaagaataaacaaggactcaaaaaac (SEQ ID NO: 306)

For: 5'-3' = aaactgggacacttgtaatgaat (SEQ ID NO: 307)

New R 5'-3'-taaaattcacatagtgcaatggtg (SEQ ID NO: 308)

Rev 5'-3' = gttttttgagtccttgtttattctt (SEQ ID NO: 309)

#### M103 = B9.117new (463 bp) C to T at position 259

For: 5'-3' = cagtaagtgaactcacacataattcc (SEQ ID NO: 311)

Rev 5'-3' = ccagttttatttcagtttcacagc (SEO ID NO: 312)

M104 = DYS257a (288 bp) Duplicated locus. Most men have both A and G alleles at position 162, however some have only A allele. The second site at position 202 is often just C, although sometimes both C and T alleles occur.

GaacttgtcgggaggcaatGGTGACATTCATTGTGACCTTAGCCAGAGCTCACAATCAA CCATGGTGCACTGAGACTAGCTCATGCACATTCATCAGGCAGATTCAGGCAC CTGGCTGTCAGAGCTGTCAGCCTTCCTCAGTAGAGGAAAATGCTACAGTCRG CACTGGCCTGGTATCAGGAAAATAGATGCCTGCAAAAAYCCACTGTGGGACC CTAAAAGTCTTGACCTCAGGTCCCCTTTGTGCTGTCTCTGTTGTCAGGATccacta aaggaggaagtgtatca (SEQ ID NO: 313)

For: 5'-3' = gaacttgtcgggaggcaat (SEQ ID NO: 314)

#### Rev 5'-3' = tgatacacttcctcctttagtgg (SEQ ID NO: 315)

# M105 = B9.6-7a (572 bp) C to T at position 478

For: 5'-3' = gggaggcaacctaagaaag (SEQ ID NO: 317)

Rev 5'-3' = aggtgaacgcattctgtcat (SEQ ID NO: 318)

# M106 = B9.6-7b (572 bp) A to G at position 411

For: 5'-3' = gggaggcaacctaagaaag (SEQ ID NO: 320)

Rev 5'-3' = aggtgaacgcattctgtcat (SEQ ID NO: 321)

#### M107 = B9.112n (376 bp) A to G at position 298

CaaaagcactcgggttcctTGTTTCAATCCCACCTCACATACACATAAGCATCATTAACA GTACAGCGTGGGGCTCTTTATCCCATCTTGTGCACCGCTTGCCTGAGAGAATT TGCTACTGGTCCTGGGGAGCCCTGTCATATTCCCTTAGCAGGCCTGCAAAGAT CTGTGTCCATTTCTTTTCCAAAAAGTCATTTTTCTCTCAACATCCCAATCTCAT TTCCAAAACTGTCAATAAATATCAAGTTTCTTAGATTTTACTCATTTCTTAAGC CAACGTATTAACCTTCTAATTTCRTGAATGCTAATAGAAAGCATGAGACACC TATGCATCATATAAAAGTGTTTTTTATTCgttgcataagtgggagtaaag (SEQ ID NO: 322) For: 5'-3' = caaaagcactcgggttcct (SEQ ID NO: 323)

Rev 5'-3' = ctttactcccacttatgcaacg (SEQ ID NO: 324)

#### M108 = B9.113n (321 bp)T to C at position 40. Probably recurrent

AGAAGAATCCGGCTGGAGACTTTAGTGATCTGGGTAGAAGATAAAATCATCC ACAATATTTACTGGGGTTTTTTTTTGCATTTCCTGAATTTGAATCTTGGCCAGAG TAAAGGGAAATATTCATCCCTCCTCCTTTTTAGCACCCATTCCCACTTAAAGC CACCTCTATCACATAAAATCCTCCACATTTaccatcattcaattcatctgtgt (SEQ ID NO: 325)

For: 5'-3' = agatggagccagcagaaag (SEQ ID NO: 326)

Rev 5'-3' = acacagatgaattgaatgatggt (SEQ ID NO: 327)

M109 = G3.15 (312 bp) C to T at position 264

GggtatcaaatgtcttcaacctAAAGTACAAGGAATTATTTCTCAGTGTTTGGAATGACTT GACTTCCTTGAAAATATTGTTGCAGAGTTGGGGACTACTTTTAAAATATCCTC CATTGAATGTAATTCTACATGAAAGCTTGATTTTTCAAGTGCAAAATGCAAGT GAGAAATAAGGCATATCATTCATTAAACCCTAATTCCAGCACTTTTAAATGA GCTACTTTCTTGTATAATATTTTAGCTATTAAGGAACAAATTGTYGCTTAAGA AATGTATCTATCATTAAAAAATgcaagtagcaggaaattccc (SEQ ID NO: 328)

For: 5'-3' = gggtatcaaatgtetteaacet (SEQ ID NO: 329)

Rev 5'-3' = gggaatttcctgctacttgc (SEQ ID NO: 330)

M110 = B9.86n (389 bp) **T to C** at position 241

For: 5'-3' = cagggaaggaccgtaaaagg (SEQ ID NO: 332)

Rev 5'-3' = atgtttatcatgtgcagtaaaggtt (SEQ ID NO: 333)

M111 = G3.19 (393 bp) -2bp (TT) deletion at position 188-189 interval. Polymorphic STS = 391 bp.

For: 5'-3' = aatettetgeaaagggttee (SEQ ID NO: 335)

Rev 5'-3' = cagctacaaaacaaaatactggac (SEQ ID NO: 336)

M112 = G3.17a (445 bp) G to A at position 286

 $A ctttttccaacagttatttttgaACTTCACTGTTACACAGTTGAGGTGACATTCATTATAAAAGAATACACAGAGGCTACTATATTAACCATTATATCTATATCTTTAGTT{\color{red} {\bf A}} ACCT{\color{blue} {\bf A}} CCT{\color{blue} {\bf$ 

For: 5'-3' = actttttccaacagttatttttga (SEQ ID NO: 338) Rev 5'-3' = tatatttcttgatgatgagaccaat (SEQ ID NO: 339)

# M113 = G3.17b (445 bp) A to G at position 112

For: 5'-3' = actttttccaacagttatttttga (SEQ ID NO: 341)

Rev 5'-3' = tatatttcttgatgatgagaccaat (SEQ ID NO: 342)

#### M114 = G3.23 (434 bp) T to C at position 387

For: 5'-3' = ttaccacacagttgagtagttctaaa (SEQ ID NO: 344)

Rev 5'-3' = caaataaaattggagcggtta (SEQ ID NO: 345)

#### M115 = G3.22 (413 bp) C to T at position 201

agtttacagtcacatcaatttggaAAGTCATACAAATATTGTCAAAAAAACTGATCTGAATCA
AATATGCCATGCTTGTTTCTTAATCCATTGAAGTTTTACTTATCATTTAAATGA
CTTGACAATATTAGTCAGTTTATATTTTCTTTTATGTAGATATTATGGGCTCCA
GAGTTTAAATTAGTATTTGATTTCACATTAYGAAACCATTATAAAAAAAGTCTC
AAATTAAGATAATTTAAGGTGATGAACACACAAACGTACACTTTGAAAGGAG
AAGGCAATGAAAACATGCATTCCAATAAAGGGGGAAAATGAGGCTGATGTG
CAACATAGTTGGGGAAATTGGTAAGAAGCTTTCTGTTACCACACAGTTGAGT
AGTTCTAAAAaaacagagatatggtagaaaaagga (SEQ ID NO: 346)

For: 5'-3' = agtttacagtcacatcaatttgga (SEQ ID NO: 347)

Rev 5'-3' = tcctttttctaccatatctctgttt (SEQ ID NO: 348)

M116 = G3.25a (429 bp) Three alleles. A to T (M116.2) or A to C (M116.1) at position 176

For: 5'-3' = aagtatgacttatgaagtacgaagaaa (SEQ ID NO: 350)

Rev 5'-3' = attcagttagattttacaatgagca (SEQ ID NO: 351)

Rev 5'-3' = attcagttagattttacaatgagca (SEQ ID NO: 354)

M118 = G3.29 (478 bp) A to T at position 109

For: 5'-3' = attetaagttteaetteetgatee (SEQ ID NO: 356) Rev 5'-3' = tagtteeetaaattaegetaeete (SEQ ID NO: 357)

M119 = G3.32 (330 bp) A to C at position 224

ATTTT**M**GCCTGTATGCTGAATTGGAATAACCCATAACATTTTCTACATCTA ATTTTAAAAAACGGTTTAAATTTGTATTAATTaagaatacatcttgtatattgtgtgaa (SEQ ID NO: 358)

For: 5'-3' = gaatgettatgaatttcccaga (SEQ ID NO: 359)

Rev 5'-3': ttcacacaatatacaagatgtattctt (SEQ ID NO: 360)

M120 = B9.87b (495 bp) T to C at position 224

For: 5'-3' = gagettggactttaggacgg (SEQ ID NO: 362)

Rev 5'-3': aaactttaaggcacttctggc (SEQ ID NO: 363)

M121 = B9.87c (495 bp) 5 bp deletion at position interval 183-187

For: 5'-3' = gagettggactttaggacgg (SEQ ID NO: 365)

Rev 5'-3' = aaactttaaggcacttctggc (SEQ ID NO: 366)

M122= G3.27a (393 bp) T to C substitution at position 73

TggtaaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGA
TACTAATTCAYGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACA
CAGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCAGCATCG
CCTGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGC
AAAAAACTATGGGGGGAACAGGGAAGTCGGTTTAATAATACTGAGTTTGTGC
AACCTCAACTTTGCTTTATAGGAAAGCAAAATCTCAATATGATAAAGTTTTCT
TCAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaag
aaaatctaattcgctg (SEQ ID NO: 367)

For: 5'-3' = tggtaaactctacttagttgccttt (SEQ ID NO: 368)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 369)

M123 = G3.27b (393 bp) G to A at position 161

TggtaaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGA
TACTAATTCATGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACAC
AGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCARCATCGC
CTGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCA
AAAAACTATGGGGGGAACAGGGAAGTCGGTTTAATAATACTGAGTTTGTGCA
ACCTCAACTTTGCTTTATAGGAAAAGCAAAATCTCAATATGATAAAGTTTTCTT
CAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaaga
aaatctaattcgctg (SEQ ID NO: 370)

For: 5'-3' = tggtaaactctacttagttgccttt (SEQ ID NO: 371)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 372)

M124 = G3.27c (393 bp) C to T at position 246

TggtaaactetacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGA
TACTAATTCATGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACAC
AGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCAGCATCGC
CTGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCA
AAAAACTATGGGGGGAACAGGGAAGTYGGTTTAATAATACTGAGTTTGTGC
AACCTCAACTTTGCTTTATAGGAAAGCAAAATCTCAATATGATAAAAGTTTTCT
TCAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaag
aaaatctaattcgctg (SEQ ID NO: 373)

For: 5'-3' = tggtaaactctacttagttgccttt\_(SEQ ID NO: 374)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 375)

M125 = B9.108a (367 bp) T to C at position 301

GccacctcttatgcctctGGCCTTTACAAAGACAGCTGGTAAGAGGCTGCCCAGCTCAT CTGAAGTACAGGATAAGATTGTCTGACTTGGAGATACCATTTTCCACTTAGCA GCCATGTAATCTTTCATATTCATTTTTTCTAAGTGGCACTTTTCTCAGATGTAA AATGGGGATAATGAGTTTATTCATCTTTGAGTTGCTCCCAAGCAGAAGTCAAC TTGAGACTATAAACTTGTGCTCACTGCAGTGCTTGAAACCGAGTTTGTACTTA ATAAATAGCTGCATACATCTTTTTCTAYACATGTCAGATGCTTAATTGTGTTT CCCGAAGATGTTGCCAAGCCgggtcctcacataactcctga (SEQ ID NO: 376)

For: 5'-3' = gccacctcttatgcctct (SEQ ID NO: 377)

Rev 5'-3' = tcaggagttatgtgaggaccc (SEQ ID NO: 378)

M126 = B9,108b (367 bp nominal) 4 bp deletion (AATA) at interval 277-280.

GccaccctcttatgcctctGGCCTTTACAAAGACAGCTGGTAAGAGGCTGCCCAGCTCAT CTGAAGTACAGGATAAGATTGTCTGACTTGGAGATACCATTTTCCACTTAGCA GCCATGTAATCTTTCATATTCATTTTTTCTAAGTGGCACTTTTCTCAGATGTAA AATGGGGATAATGAGTTTATTCATCTTTGAGTTGCTCCCAAGCAGAAGTCAAC TTGAGACTATAAACTTGTGCTCACTGCAGTGCTTGAAACCGAGTTTGTACTTA ATAAATAGCTGCATACATCTTTTTCTATACATGTCAGATGCTTAATTGTGTTT CCCGAAGATGTTGCCAAGCCgggtcctcacataactcctga (SEQ ID NO: 379)

For: 5'-3' = gccaccctcttatgcctct (SEQ ID NO: 380)

Rev 5'-3' = tcaggagttatgtgaggaccc (SEQ ID NO: 381)

M127 = G3.30 (412 bp) C to A at position 372 bp

TgaaaggaaatcagtgtaagagcTAGAGGTAGCGTAATTTAGGGAACTAATCAGGAAAGA GGTATTAACATTTCTGAATCCTTAGTTTCACTTATCCTTTCAATTCACAAGATT GCTTTATTTCACATTTTGATAAAGACCAAAATGGTCCAAAAATAAGGGGAGG AAGAACCTATACTACAAGAACCGAATTCCCAGACACTCAGGATAAACTTTAG GTATATCCTTCAATCAGCTTTGTTCCAAATACAGGTAACGAGCCAGGCAATGT TACGGAAAATAAGGGTAAGATAAAGCAAATATCCTGTGCTTTGGTTAACAAA CAAAACTGTATCACAAGTCAAACTCGTACAAAAGGCAGGAGAAGAGGT**M**TG GAAGATCTGTTAGGtgctgaactacagtcacctttaca (SEQ ID NO: 382)

For: 5'-3' = tgaaaggaaatcagtgtaagagc (SEQ ID NO: 383)

Rev 5'-3' = tgtaaaggtgactgtagttcagca (SEQ ID NO: 384)

For: 5'-3' = actttttccaacagttatttttga (SEQ ID NO: 386) Rev 5'-3' = tatatttcttgatgatgagaccaat (SEQ ID NO: 387)

M129 = A8.04 (255 bp) G to A at position 221.

For: 5'-3' = aatggcttactacaaagaacatttc (SEQ ID NO: 389)

Rev 5'-3' = tacacggtctctaccaaagaaga (SEQ ID NO: 390)

M131 = A8.14n (306 bp) 9 bp deletion at interval 93 to 101

CacacccagaatacaataattttAAAAACATAATAAAGGTCAATTTAGAGCAGAGAAATTA TTCTTTTAAATTACAAATGTTTGCTGTTCAG**GCAAATTAC**ACACAGAAAGTTA AGAATAACCCTTTAAATGATAGGAAAAGGCATTAGTAAGATAAAATGTGATT ACTATTGAGATAAATATTTGCTATAAAAATAATTCAATTTGGTTAAACACAAA TTGACTTCTTAAATAATCTTAAACATTAAGTAGAAGTAATTTTAGCTTATCAG TAAATTTGAgaaaatgtacacttgtagaataaaaaag (SEQ ID NO: 391)

For: 5'-3' = cacaccagaatacaataatttt (SEQ ID NO: 392)

Rev 5'-3' = ctttttattctacaagtgtacattttc (SEQ ID NO: 393)

M132 = B9.67b (568 bp) G to T at position 482

For: 5'-3' = aacagaattatcaggaaaaggttt (SEQ ID NO: 395)

Rev 5'-3' = ttttacttgttcgtgtactttcaa (SEQ ID NO: 396)

For: 5'-3' = tgaaatggaaatcaataaactcagt (SEQ ID NO: 398)

Rev 5'-3' = ccttttctttttctttaacccttc (SEQ ID NO: 399)

For: 5'-3' = agaatcatcaaacccagaagg (SEQ ID NO: 401)

Rev 5'-3' =tctttggcttctctttgaacag (SEQ ID NO: 402)

M135 = A8.08F-newR (211 bp nominal vs 212) 1 bp insertion (+ C) at position 150 = site c, within homopolymer A track.

Site a (A)<sub>10</sub> -TTT most males

Site c (A)9CATTT = M135

Site d (A)11TTT

For: 5'-3' = tgaaatggaaatcaataaactcagt (SEQ ID NO: 404)

Rev 5'-3' = cettttetttttetttaaceette (SEQ ID NO: 405)

M136 = B9.61 (339 bp) C to T at position 196

AtgtgaagacaacactgtgtggGAGAACCTAGGAAAGTAATTTTACATGCTAAAATGAGT
TTCCCTAGTTAATGTTAACATGAACTACCAACCGTATTACCTTCTCCTCAGGA
GATAAGTTTTGTTTGCTATTGCTGACAGGAAAGCCACTGCCAAATTCTTTGGA
ATGAATATCAGCTCCATATTCAACTGTCAYGTCTCTCAATGCTGCTCACCA
GCCTCCAGAATTCCTTCTCTACAAGTTCTGTAGGCACCATCTGTGAAAACACA
TGTAAAAGGTTATCATAGCCCACTATACTTTGGACTCATGTCTCcatgagaactaagac
taccacaa (SEO ID NO: 406)

For: 5'-3' = atgtgaagacaacactgtgtgg (SEQ ID NO: 407)

Rev 5'-3' = ttgtggtagtcttagttctcatgg (SEQ ID NO: 408)

M137 = G3.27d (393 bp) **T to C** at position 289

TggtaaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGA
TACTAATTCATGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACAC
AGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCAGCATCGC
CTGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCA
AAAAACTATGGGGGGAACAGGGAAGTCGGTTTAATAATACTGAGTTTGTGCA
ACCTCAACTTTGCTTTAYAGGAAAGCAAAATCTCAATATGATAAAGTTTTCTT
CAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaaga
aaatctaattcgctg (SEQ ID NO: 409)

For: 5'-3' = tggtaaactctacttagttgccttt (SEQ ID NO: 410)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 411)

M138 = A8.17(442 bp) C to T at position 291

AacttccaaaactgtgaaaagattGTTTTTAAAAGGCTATAACAGTGACTTTCAGGTGAAGA CTTGGACAAATAGATAATTTCTGTACCCATTAAAATCAGGGGCTGTTACTATG TTTGAAGACATTGTCGCCACAGCTTGAAGTCTGTAAGGAAAACCTGTAAAAT TAGTGGGTGCCCACTCTAGTTTTAATCATTTGAGTTTCCACTCCTCATTGTGGT TGAACTATTTTATAACTCTGCAAAAATCTAGAAAGTTGAAAAGAAACCAAAGA TACTTTCCCTTTTCTTCYCACCTTCCTACCCTTGGCCCACCTCCTTCTCCACC TACTACTCCACATGGAACCTGGAGATTTGAGTCGGGGAGTGATGTAATACCT GCGGCGCGTTGGCCCTTTACACACCTGTCAGCCATTTCAAGGCctgaaggggctgcttt aatc (SEQ ID NO: 412)

For: 5'-3' = aacttccaaaactgtgaaaagatt (SEQ ID NO: 413)

Rev: 5'-3' = gattaaagcagcccttcag (SEQ ID NO: 414)

M139 = A8.28a (459 bp nominal vs 460) 1 bp deletion at position 401. 5 G's to 4 G's.

TtactgataatgccatattgttttgGCTTAATATCAGGCTAAGTAACCACAGTATTCTGATTTA **AAAAAAA**CATACTAGAGAGCAAGTTTATTGACAAATCTTTAGGAACTTCAG
GTACAGCATATGATTTCTGAACTATGTGTGTAAATAAGGTTTTGTTTATTCAA
ATTTAACACAGGGTAGTCTGTGTATGCCTTCCGATTTGATAGCTCTAATAAAA
CACTTTAATAGTACCATATCAAAATAAATTTTATCATCATCGATTTTCTTCTTAA
TATGAAATAACACATATTTGTGATTTTTCTAAGAGTCAAAAATCA
TTTTAGGTATAAAATATACCCCGAAAGTTTTATTTTTTTCATTTTTATAATTAA

TCTGACTTGGAAAGGGGAAAAAAGCTCAAAGGGTATGTGAACATTTCATT AAGATaggaccattggtgtctgagaa (SEQ ID NO: 415)

For: 5'-3' = ttactgataatgccatattgttttg (SEQ ID NO: 416)

Rev 5'-3' = ttctcagacaccaatggtcct (SEQ ID NO: 417)

M140 = A8.28b (459 bp nominal vs 460) 1 bp insertion within 9 A's homopolymer (most men) to 11 A's at position 73. Recurrent because 11 A's found in different haplogroups.

For: 5'-3' = ttactgataatgccatattgttttg (SEQ ID NO: 419)

Rev 5'-3' = ttctcagacaccaatggtcct (SEQ ID NO: 420)

M141 = A8.30a (424 bp nominal) T to A at position 51. Locus also has two homopolymer T tracks which are both polymorphic. See next below.

CatcttaaaatacatttcatagctttTCAAACTCAAATATGAAAACAATT**W**GTTTTTTAGA**TT TTTTTTT**CTTTTACTTCAAGTTCTTTATATTCTAGACTAACACTTTAGGGCA
GATATTGGAGGGTGTGTCTCTCTTGGTGCAACTATTGCCTTTGCTTCAAATGG
TGGCATATGGAGGAGGACACAACCTGTAGGAAGTGTTCAAGGAGTCTGGTAG
TGACACCTGCTCAATATTGCTAGTGATAAAACTGTAGCCACTGTATAGCAATA
TCTGCCTGTAGAATGTCATTTCCTTTGAGGGGTACA**TTTTTTTT**AGAGTTTCC
TATAACCTCTAGAGCTGAACTTCATAAAAAATAGGTAAAGGTTGGCCTTAAAA
AGCCTACATTACACACTTTCaggatgctagacctaatagtaagc (SEQ ID NO: 421)

For: 5'-3' = catettaaaatacatttcatagettt (SEQ ID NO: 422)

Rev 5'-3' = gcttactattaggtctagcatcct (SEQ ID NO: 423)

For: 5'-3' = catcttaaaatacatttcatagcttt (<u>SEQ ID NO: 425)</u>

Rev 5'-3' = gcttactattaggtctagcatcct (SEO ID NO: 426)

#### M143 = B9.50b (385 bp) G to T at position 246

For: 5'-3' = atgetataataactaggtgttgaag (SEQ ID NO: 428)

Rev 5'-3' = aattcagcttttaccacttctgaa (SEQ ID NO: 429)

#### M144 = B9.99 (452 bp) T to C at position 342

For: 5'-3' = agcacaagggtcacattgag (SEQ ID NO: 431)

Rev 5'-3' = aggacaaggctttttgttgtt (SEQ ID NO: 432)

#### M145 = A8.05b (208 bp) G to A at position 166

RAGAGCCAGCCTTAGCCTAATCaagaaccatgatccaaaaagg (SEQ ID NO: 433)

For: 5'-3' = ttcagcaagagtaagcaagagg (SEQ ID NO: 434)

Rev 5'-3' = cctttttggatcatggttctt (SEQ ID NO: 435)

#### M146 = G3.04d (395 bp) A to C at position 141; has(GTTTT)6 motif

For: 5'-3' = gaatggggtgttacatggaga (SEQ ID NO: 437)

Rev 5'-3' = cctattatgagagacctgttttcc (SEQ ID NO: 438)

M147 = G3.35 (439 bp nominal) 1 bp insertion (extra T). Associated with GTTT repeat. 3 T's to 4 T's at position 116. Locus also has T homopolymer which cause stutter bands during PCR.

For: 5'-3' = gtattctggggcaattttagg (SEQ ID NO: 440) Rev 5'-3' = ttgatacaagaggttattttaagca (SEQ ID NO: 441)

M147new = G3.35 (276 bp nominal) 1 bp insertion (extra T). Associated with GTTT repeat. 3 T's to 4 T's at position 97.

#### M148 = B9.67c (568 bp) A to G at position 314

For: 5'-3' = aacagaattatcaggaaaaggttt (SEQ ID NO: 444)

Rev 5'-3' = ttttacttgttcgtgtactttcaa (SEQ ID NO: 445)

#### M149 = B9.67d (568 bp) G to A at position 469

AacagaattatcaggaaaaggtttCATAAAAATAAAAATCTTTTTAAACTTATGAAAGATGCT CAATATAAAAAACTGTAAACCAGGGAAATGCAAATAAAAATTACAATGAAA TACTACACACCTCCCAGAATGGCTAAAATGAAAACAAAACTGTCAATTCTAA GTGTTAGTGAGGACATGTGGTAACCAGAACTGGCATCCAATACTAGCTGATA AACTCGTCAATCATTTGTAAAAACAGTCTGACAATAATCCACTAGTGAAAAT ATACATAGTCTCAGTCACAATTCTATCCTGTCTATCTAGGTAACAGAAAT

GTCTACATACGTTACCTAGAAACATATACTTTAATATCCACAGAATTACTTGA AATAGCCAAAAATTGGTAACTACCAAAAGTTGAATGGTAAAACAGATAGAA AAAAAGCTATGCCTAACAAAACTACACTTAATAGAACAAAGCAGCRTGAGCAT TAATAGAACCATATAAATGCATTTTTTGAACCACTAAAAGAAGAAGCCAATA CAAAAGAGGTGATTAAttgaaagtacacgaacaagtaaaa (SEQ ID NO: 446)

For: 5'-3' = aacagaattatcaggaaaaggttt (SEQ ID NO: 447)

Rev 5'-3' = ttttacttgttcgtgtactttcaa (SEQ ID NO: 448)

M150 = B9.18 (289 bp) C to T at position 146

For: 5'-3' = gcagtggagatgaagtgagac (SEQ ID NO: 450)

Rev 5'-3' = cctactttcccctcttctg (SEQ ID NO: 451)

M151 = B9.58b (422bp) G to A at position 209.

ActtaatttatagtttcaatccctcaGTAATTTTAACTTACTTCTATTTTAAGAACTATAACCA AACTATCTGTAAGACTTTTAAGCACTATCATACTCAGCTACACACATCTCTTAAC AAAAGAGGTAAATTTTGTCCTTTTTTGAACGTCATAGAGTATACTCACACAAA CCAAGAAGAAACAATCTACTACATACCTACGCTATATGRTATATAACTATTG CTCCTAGGCTACAAATTAGTGCGACACTATTGTACTGAATATTATAGGCCATG TAACACAATGGTTTAAGTATCTGTGCCTCTAAACACAGAAAAGATATAGTGA AAGTACAGTATTGCTCCTTTATTAAACTCAAAATGTTATGCAGCATATGACCG ACTATAAAATAGCGCTTATCCCCGTTATCCCCTCTAACCCCG (SEQ ID NO: 452)

For: 5'-3' = acttaatttatagtttcaatccctca (SEQ ID NO: 453)

Rev 5'-3' = ttcatggagatgtctgtatctgg (SEQ ID NO: 454)

M152 = B9.13 (287 bp) C to T at position 101

AagctattttggtttcctttcaAGAAAGGGCTGTGGTCTGTGGAAGGTGTCAGGAACATATT TTCCACGGTCTGCTTCTCTGATAATGTTCTTCTYGGCCCACCTGAGAC ATAATCCCTGAGCTCCGAGCCCTTTTTGACTGAAGCTCCTGTTGAACAAGATT CTCAACGTTTCTACCCTGATCCACCTTCTGCCGCCGCCGCCGTCGCCTCTCCAGAG CCCGGCTCCTTGTCCGACTCCCTTGATGTTCAAATTTTTCCAGCTGcaatcatacccac acaaggc (SEQ ID NO: 455)

For: 5'-3' = aagetattttggtttcctttca (SEQ ID NO: 456)

Rev 5'-3' = gccttgtgtgggtatgattg (SEQ ID NO: 457)

M153 = A8.28c (459 bp nominal) T to A at position 427 bp

For: 5'-3' = ttactgataatgccatattgttttg (SEQ ID NO: 459)

Rev 5'-3' =ttctcagacaccaatggtcct (SEQ ID NO: 460)

M154 = B9.58c (422bp) T to C at position 252.

For: 5'-3' = acttaatttatagtttcaatccctca (SEQ ID NO: 462)

Rev 5'-3' = ttcatggagatgtctgtatctgg (SEQ ID NO: 463)

M155 = G10.57c (327 bp) G to A at position 251

For: 5'-3' = tetetaacttetgtgageeae (SEQ ID NO: 465)

Rev 5'-3' = ggaaaaactaaactctaaatctct (SEQ ID NO: 466)

M156 = A8.05c (208 bp) A to G at position 147. Linked to M145 derived allele. TtcagcaagagtaagcaagaggCACTGAGCCGCTGGAGTCTGCACATTGATAAATTTACT TACAGTCGTAAATAAATTGCATCATCTTCAGCTAGTAACACAGAGTCTAATTT TTATAGCGGCATACTTGCCTCCACGACTTTCCTRGACACCAGAAAGAAAGGC GAGAGCCAGCCTTAGCCTAATCaagaaccatgatccaaaaagg (SEQ ID NO: 467)

For: 5'-3' = ttcagcaagagtaagcaagagg (SEQ ID NO: 468)

Rev 5'-3' = cctttttggatcatggttctt (SEQ ID NO: 469)

M157 = B9.12b (352 bp) A to C at position 176

For: 5'-3' = getggcaagacacttetga (SEQ ID NO: 471) Rev 5'-3' = aatatgtteetgacacettee (SEQ ID NO: 472)

For: 5'-3' = tgaaatggaaatcaataaactcagt (SEQ ID NO: 474)

Rev: 5'-3' = ccttttctttttctttaacccttc (SEQ ID NO: 475)

M159 = G10.83 new b (190 bp) A to C at position 89

For: 5'-3' = attggattgatttcagccttc (SEQ ID NO: 477)

Rev 5'-3' = attttattttctgtgttccttgc (SEQ ID NO: 478)

#### M160 = B9.47b (361 bp) A to C at position 251

CagaataataggagaatttttggtCAAATAAAAGGCCATATTATATTTCTTTTGATAAAAGT ATCATGTGTTCAGTATGTTTTATTTTTGAAAATAATTAACATGACAGGAATAT ATTTGAAAAAAAATTCCAAAAAAAAGCTAAATATACAAACTAAGAAAATTATAT GATTATACTTATCTGCAGTATTGTAAAACAATAGTTCCAAAAAACTTCTGAATT ACAAGTTTAATACATACAACTTCAATTTTC**M**ACTACATT**GT**GGTTAGACGTT CAGAGGAATCACAAAGGACCTCAACATGCTAGATAAGAAAATGTATTTTTA AATGTTTTGGCTCAgctgcttagaaaataaggaaaat (SEQ ID NO: 479)

For: 5'-3' = cagaataataggagaatttttggt (SEQ ID NO: 480)

Rev 5'-3' = attttccttattttctaagcagc (SEQ ID NO: 481)

#### M161 = A8.05d original (460 bp) C to A at position 111

For: 5'-3' = tcacagcagcttcagcaaa (SEQ ID NO: 483)

Rev: 5'-3' = cctttttggatcatggttctt (SEQ ID NO: 484)

new R 5'ataaaaattagactctgtgttactagc (SEQ ID NO: 485) (used with F primer, just amplifies the first 2 sites including homopolymer T region.

M162 = DYS257b (288 bp) =

C/T at position 202), most men are just C at position 202

Duplicated locus. Most men have both A and G alleles at position 162, however some have only the A allele. The second site at position 202 is often just C, although sometimes both C and T alleles occur on a chromosome background that is both A and G at position 162.

GaacttgtcgggaggcaatGGTGACATTCATTGTGACCTTAGCCAGAGCTCACAATCAA CCATGGTGCACTGAGACTAGCTCATGCACATTCATCAGGCAGATTCAGGCAC CTGGCTGTCAGAGCTGTCAGCCTTCCTCAGTAGAGGAAAATGCTACAGTCRG CACTGGCCTGGTATCAGGAAAATAGATGCCTGCAAAAAYCCACTGTGGGACC CTAAAAGTCTTGACCTCAGGTCCCCTTTGTGCTGTCTCTGTTGTCAGGATccacta aaggaggaagtgtatca (SEO ID NO: 486)

For: 5'-3' = gaacttgtcgggaggcaat (SEQ ID NO: 487)

Rev 5'-3' = tgatacacttcctcctttagtgg (SEQ ID NO: 488)

#### M163 (340 bp) G10.35b A to C substitution at position 168

For: 5'-3' = gcagcatataaaactttcagg (SEQ ID NO: 490)

Rev 5'-3' = aaaacctaactttgctcaagc (SEQ ID NO: 491)

#### M164 = G10.100b (493 bp) T to C at position 329

TagaagtagcagattgggaggaCATGTGTTCAAGTTGTACTACTTGTATGTCTTGTTTA
GATATTACAGTCTTTTTCTTTTATCAGAAAAATAATTGAATAATGATAAAATCA
GTTGCAGATTAAGACAGATTATCTGTTGCAGTCTTCTCAAAACTTAATTTAAG
TACATTATTTTCAGCTAGCATTTCTTCCTTCACATAGAACCTCCATGTGTGGA
GGGATTTCCTAATGAGTCTATTGTATGTACAATAGCACTTAATGACATAGCTT
TTAAATAATAACAGGATTTTACCAAATGTTTAATATGTGCCAGGCATCAAGC
ACCYTACACAGTTTAATTATTGCATAGATTTGGACAGCAACTCTGCAAGTTA
GGTATGGTCATGAACCTTTGCAGATAAGGAAACTGTGTTTCACAAGGAAAG
AAATTGTCCTGGATCATACAATAAGCTAGGATTTGCTCCAgaccatttttttcattttatcagg
(SEO ID NO: 492)

For: 5'-3' = tagaagtagcagattgggagagg (SEQ ID NO: 493)

Rev 5'-3' = cctgataaaatgaaaaaatggtc (SEQ ID NO: 494)

#### M165 = B9.008c. (340 bp) A to G at position 132.

AaagcgagagattcaatccagGATGACAGAATGCGTTCACCTTTAAAGGGATTAAAAGA AGTATAATACAGTCTGTATTATTAGATCACCCAGAGACACACAAAACAAGAA CCGTSAATTGAATTAGTGGTATACTAATAGAGTGGTTTTACCTGAAATATTTA CACATCAATCCTACTGAATTCTTACAACAAATGATTTAGATTAGCTATTGTAT TCACCAGTTGAAAGAACAGAAAATATTGAGGGAGATAACTTGTGTCAGTGCA ACTTAATCAGATTTAGGACACAAAAGCAACTACATAATGAAAAAGAGAgctggt gacttaacttgctaaaa (SEQ ID NO: 495)

For: 5'-3' = aaagcgagagattcaatccag (SEQ ID NO: 496)

Rev 5'-3' = ttttagcaagttaagtcaccagc (SEQ ID NO: 497)

#### M166 = G3.27e (393 bp) G to A at position 53

tggtaaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAA**R**CAATTGAGA TACTAATTCA**T**GCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACAC AGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCA**G**CATCGC CTGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCA AAAAACTATGGGGGGAACAGGGAAGTC**G**GTTTAATAATACTGAGTTTGTGCA ACCTCAACTTTGCTTTATAGGAAAGCAAAATCTCAATATGATAA**A**GTTTTCTT CAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaaga aaatctaattcgctg (SEQ ID NO: 498)

For: 5'-3' = tggtaaactctacttagttgccttt (SEQ ID NO: 499)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 500)

#### M168 = DFFRY Ex01B site a(473 bp) C to T at position 371 noncoding

AgtttgaggtagaatactgtttgctGGTCTTAAAAACTGTGGTATTTTGGTGATTCCATAAAT TAGGTCAGATACTTCCACTGGAGGGAAACAGTTTAAAGGATATTTGTGATAC TATTAATAGAATGAGGAAGACACACCAGATATTTAGGAGGGAATTAGCGAGC TTGAAACTAAGAGCTGGTTTGAATGAGGACTGGGTCATAAGTGATTTCAAGTA CCAGATTAAGGCACTGAGATTTTTTTTTAAGCACTGAAGTCAGATTTTTCC TTTTAAAAGAAAGGATTCATGATGAAAATCTGCTTTTTGTTTTTGCAGAGAGCTT GGAGATAATTCTGGTGGCTGTGTGGAGTATGTGTTGGAGGTGAGTYGCTAGC TGAAGAATTAAAACAATAGTTTTAGCAGTTTTGGGTAAGAGATTTTACAGAA ATGTTTTGTGGAAAAAACAGATAAAACtgaacagtcagagacctatgagatt (SEQ ID NO: 501)

For: 5'-3' = agtttgaggtagaatactgtttgct (SEQ ID NO: 502)

Rev: 5'-3'= aatctcataggtctctgactgttc (SEQ ID NO: 503)

#### M169 = DFFRY Ex01B siteb (473 bp) T to C at position 97 noncoding

For: 5'-3' = agtttgaggtagaatactgtttgct (SEQ ID NO: 505)

Rev: 5'-3'= ccagggcccgagggactctt (SEQ ID NO: 506)

#### M170 = DFFRY Exon08 (405 bp) A to C at position 327

For: 5'-3' = tgcttcacacaaatgcgttt (SEQ ID NO: 508)

Rev 5'-3' = ccaattactttcaacatttaagacc-3' (SEQ ID NO: 509)

M171 = DFFRY Ex01B sitec (473 bp) G to C at position 440 noncoding?

For: 5'-3' = agtttgaggtagaatactgtttgct (SEQ ID NO: 511)

Rev: 5'-3'= ccagggcccgagggactctt (SEQ ID NO: 512)

M172 = DFFRY Ex45 (345 bp) T to G at position 197

For: 5'-3' = ttgaagttacttttataatctaatgctt (SEQ ID NO: 514)

Rev: 5'-3'= ataatttattactttacagtcacagtgg (SEQ ID NO: 515)

For: 5'-3' = aagaaatgttgaactgaaagttgat (SEQ ID NO: 517)

Rev: 5'-3'= aggtgtatctggcatccgtta (SEQ ID NO: 518)

M174 = DffryEx38 (348 bp) T to C at position 219

AcatctcagatcgttgtttggtTCATAAAAATCTGTTTCTTCCATGTACCAAGCAAAATAAA CACATCACTAAAATTTGACGTTCATAGATGTTTCTGTTTTAGGTATGATGCAC TGTGCGTTCTTCCGTCACAGCAAAAATGTACGTTTTTGGTTTACTCATAAT GTCCTTTTTAATGTATCAAATCGCTTCTCTGAATACCTTCTGGAGTGCCCYAG TGCAGAAGTGAGGGGTGCATTTGCAAAACTTATAGTGTTTATTGCACACTTTT CCTTGCAAGATGGGTCTTGTCCTTCTCCTTTTTGCATCTCCAGGACCTTCTAGTc aggtaattgcatggcttttt (SEQ ID NO: 519)

For: 5'-3' = acateteagategttgtttggt (SEQ ID NO: 520) Rev: 5'-3'= aaaaagccatgcaattacetg (SEQ ID NO: 521)

For: 5'-3' = ttgagcaagaaaatagtaccca (SEQ ID NO: 523) Rev: 5'-3'= ctccattcttaactatctcaggga (SEQ ID NO: 524)

#### M178 = G10.72b (514 bp) C to T at position 220

For: 5'-3' = taagcctaaagagcagtcagag (SEQ ID NO: 526) Rev 5'-3' = cagagggagcaatgaggaca (SEQ ID NO: 527)

#### M179 = Dffry exon 07 (426 bp) C to T at position 316

For: 5'-3' = acactactgtgctgtaatttgtgaa (SEQ ID NO: 529) Rev 5'-3' = agacaggttcacatcactaatgg (SEQ ID NO: 530)

M180 = Dffry exon 11(447 bp) T to C at position 402

For: 5'-3' = acactactgtgctgtaatttgtgaa (SEQ ID NO: 532) Rev 5'-3' = tggtaagatttctctacatgaacag (SEQ ID NO: 533)

M180 = Dffry exon 11(232 bp) T to C at position 128

CaggaccttgataatactgGGCAGCACAGGTAAGAAAGTGAGATGATAGCTATTTTCTA AGAAAGATACCAAAAAGGAGAAAATTTTTGGTAACCCTTATATAATGGCCAG CAATTTAGTATTGCCYGACTTTTACTAATGCATGTGctgttcatgtagagaaatcttaccaAG AATTTTTAAACAAAAAAAAATAACATTTTTCTGTCTTTgtatatatattcatggtagcaa (SEQ ID NO: 534)

NEW F 5'-3' = caggaccttgataatatctg (SEQ ID NO: 535) NEW Rev 5'-3' = ttgctaccatgaatatatatac (SEQ ID NO: 536)

**M181** = Dffry exon 12 (294 bp) **T to C** at position 130

GcttttatttattctacttttgtttttTCAACAGGCAGGAAAACATGAAGCCATTGTGAAGAATG TACATGATCTGCTAGCAAAGTTGGCTTGGGATTTTTCTCCTGGACAACTTGAT CATCTTTTTGAYTGCTTAAGGTAGTAGCTTGAATAGTAAAGTATTGCCAAAT AGTAAATATTGCCAGTTAATTCTAAGTAAAGTTTAATTCGTTAGATTTCTTTT GCTTATAGCTAGTGTGCTTAACTAACATTTTCATGGAAGAATCTCTGatgaaaaaga attggtcattgtt (SEQ ID NO: 537)

For: 5'-3' = gcttttatttattctacttttgttttt (SEQ ID NO: 538) Rev 5'-3' = aacaatgaccaattcttttcat (SEQ ID NO: 539)

M182 = Dffry exon 13 (364 bp) C to T at position 38

TattcaaagacttaaagcagtggttaATGTAAACAAAYGTAATAAATTATGTGGTATTTATA
TCATTTAAATACTTTCTTTAGGCAAGTTGGACAAATGCAAGTAAAAAGCAAC
GTGAAAAGCTCCTTGAGTTGATACGCCGTCTTGCAGAAGATGATAAAGATGG
TGTGATGGCACACAAAGTGTTGAACCTTCTTTGGAACCTGGCTCAGAGTGAT
GATGTGCCTGTAGACATCATGGACCTTGCTCTTAGTGCCCACATAAAAATACT
AGATTATAGTTGTTCCCAGGTATGGGAGTGTTTCTTTGTTCAGTTTTCTGACTT
TCCTTCACAAGTaggataacttagttacaagatgattcc (SEQ ID NO: 540)

For: 5'-3' = tattcaaagacttaaagcagtggtta (SEQ ID NO: 541) Rev 5'-3' = ggaatcatcttgtaactaagttatcct (SEQ ID NO: 542)

M183 = Dffry exon 19 (427 bp) A to C at position 324

For: 5'-3' = actgggtaaatatgactatgattgag (SEQ ID NO: 544)

Rev 5'-3' = ttccttttaacctattattactttcc (SEQ ID NO: 545)

#### M184 = Dffry exon 23 (305 bp) G to A at position 62

For: 5'-3' = cactttattttagtctgtgtctttttc (SEQ ID NO: 547)

Rev 5'-3' = aaacttagtaacatctatttctcctct (SEQ ID NO: 548)

#### M185 = Dffry exon 27 (430 bp) C to T at position 89

For: 5'-3' = ggagtacctatcactgaatgtgc (SEQ ID NO: 550)

Rev 5'-3' = gtcattcatttctgcttggaac (SEQ ID NO: 551)

M186 = Dffry exon 30 site a (365 bp nominal) -1 bp deletion (4G's to 3 G's) at position 62 (364 bp = mutant) 325 bp w/out homopolymer

TtgcatttactgttctagagagttctCAAAAAGAAATAGGAAACCACTTGAACAGTTTGGG**G** AAGTTGTATAGAAGATCTCATTTCCTTCCAGCTCTCTGTTCTCCTAACTCCTTG TCCTTTTCTATCTCCATGTTGTGAGTTGGGCCTATAATATTTTTCCTTTTGCAG GATAATGTTAAAAAACACAGGTGAAAACAGGTGTCGAAGAGCCAATACTGGAA GGCCACCTTGGGTAACAAAAGAGTTATTGGCCTTTCAAACTTCTGAGAAAA AGTATCACTTTGGTTGTGAAAAAAGGAGgtgctaatctcattaaagtaagtacTTTTTTTTCT TTTTTTGAgatggagtcttgctctgtgg (SEQ ID NO: 552)

For: 5'-3' = ttgcatttactgttctagagagttct (SEQ ID NO: 553)

Rev 5'-3' = ccacagagcaagactccatc (SEQ ID NO: 554)

newRev 5'-3'=gtacttactttaatgagattagcac (SEQ ID NO: 555) Homopolymer clipped off

M187 = Dffry exon 30 site b (366) IGNORE Homopolymer in tree T(10 to 11 T's) 325 bp w/out homopolymer

For: 5'-3' = ttgcatttactgttctagagagttct (SEQ ID NO: 557)

Rev 5'-3' = ccacagagcaagactccatc (SEQ ID NO: 558)

newRev 5'-3'=gtacttactttaatgagattagcac (SEQ ID NO: 559) Homopolymer clipped off

M188 = Dffry exon 31 (401 bp) C to T at position 185

GtattcctttgaagaaacatattgTTCCTAACCTATATTTTCTACTAATAACATGTAATGTCT
TTTTCTAACTTACTAGGAATTAATTGATGATTTCATCTTTCCCGCATCCAAAGT
TTACCTGCAGTATTTAAGAAGTGGAGAACTACCAGCTGAGCAGGCTATTCCA
GTCTGTAGTTCACCYGTTACCATCAATGCCGGTTTTGAGCTACTTGTAGCATT
AGCTATTGGCTGTGTGAGGAATCTCAAACAGATAGTAGACTGTTTGACTGAA
ATGTATTACATGGGCACAGCAATTACTAGTGAGTATTTAAATTATAAAGCTG
TTTTGTTCATTAATAATACTTCACTGTAAAATTTTATTTGGTGTTTTAgaaaaaaatta
acttgtgatggactt (SEQ ID NO: 560)

For: 5'-3' = gtattccctttgaagaaacatattg (SEQ ID NO: 561)

Rev 5'-3' = aagtccatcacaagttaattttttc (SEQ ID NO: 562)

M189 = Dffry exon 34 (378 bp) G to T at position 191

For: 5'-3' = acteteagettatgtttgtcattg (SEQ ID NO: 564)

Rev 5'-3' = gcttttggtacactctgctttt (SEQ ID NO: 565)

M190 = Dffry exon 44 (346 bp) A to G at position 73

 $CtctgtcacaagtaaggaaatgatCGTGAAATTTTTGTATTAGCATTTTAAGCTGATACTGAAATCATTCT{\bf R} AATTCTAAATAGTTTTATTTTTTTTTCTAAAGGGTAACGGAGATCTTAAAAGAAAATGGACCTGGGCAGTGGAATGGCTAGGAGATGAACTTGAAAGAAGACCATATACTGGCAATCCTCAGTATAGTTACAACAATTGGTCTCCTCCAGTACAAAGCAATGAAACAGCAAATGGTTATTTCTTAGAAAGATCACATAGT$ 

GCTAGGATGACACTTGCAAAAGCTTGTGAACTCTGTCCAGAAGAGGTAAAAA AAaaaaaggctaccaatggacag (SEQ ID NO: 566)

For: 5'-3' = ctctgtcacaagtaaggaaatgat (SEQ ID NO: 567)

Rev 5'-3' = ctgtccattggtagccttttt (SEQ ID NO: 568)

M191 = DBY exon 2 (429 bp) T to G at position 342. Non-coding (cDNA bp# 175+120) TtgcatttgtcatggttggtTGACCTGGACATCTTTAAAATTTGGCAGGTAATACCAGGCC GACATGGCAGCTAAGTTTGTGGTACAGGATAAGATTGGAATCTAGGTCTCAT TTGTCTTTTGTGATGTTATCTGTTCTTGTGTATCAGCATGTGAGCTATTGATAT CTCTTCTAGCTTGCTAATCTGGACCTGAACTCTGAAAAACAGAGTGGAGGAG CAAGTACAGCGAGCAGTAAGTAAAACTTTTTTTAAAAAATGGAGTGTTTATCA GAGCTTAATGTTAATGTCTTACTGGACTTGTTAATTTTAAATTTACATTTTTT CTTTACAACTTGACTAKATGAAAAATATGAGATATTTTGGTGTGTCTGGGTAAT AAAATACACTGTTTACCTATGTCTGCTgaaaatacaaaaaattatcctggc (SEQ ID NO: 569)

For: 5'-3' = ttgcatttgtcatggttggt (SEQ ID NO: 570)

Rev: 5'-3'= gccaggataattttttgtattttc (SEQ ID NO: 571)

M192 = DBY STS 02 (457 bp) C to T at position 202.

For: 5'-3' = catgggctgctgacatttt (SEQ ID NO: 573)

Rev: 5'-3'= aaatccttttggtttgtttgttt (SEQ ID NO: 574)

M193 = DBY STS 03a (426 bp nominal) + 4 bp insertion (CAAA) at position 56.

For: 5'-3' = gcctggatgaggaagtgag (SEQ ID NO: 576)

Rev: 5'-3'= gccttctccatttttgacct (SEQ ID NO: 577)

M194 = DBY STS 03b (426 bp nominal) T to C at position 101.

For: 5'-3' = gcctggatgaggaagtgag (SEQ ID NO: 579) Rev: 5'-3'= gccttctccatttttgacct (SEQ ID NO: 580)

M195 = DBY STS 06 (515 bp nominal) A to G at position 430

For: 5'-3' = ccactcagctttcctcaggt (SEQ ID NO: 582) Rev: 5'-3' = cgttcgtttagtcataagatcg (SEQ ID NO: 583)

M196 = DBY STS 07 (445 bp) C to G at position 330.

TtagacaacttactactttgatgtcctGTTGGCTCAGTAATGCTCACGATACCAATTGTTTTGA CAAAATAAATTTACTAAACTTGGCCTAAAATCAAACCTTGGCACAGAGGTAT GATACAACTTTAACAGGAGTCATCAATTCATCCATAAATATAAAAAGGGAAA AAAACTTAAGGCAGTAGTCTGCATTAGGACTGTTTGAGTTTTGCAGACTTGGG GTTGGGAGAACATCTTAAAGCATTAAAGCATAAGTTTTTTGTATGGCCAACCTT ACTAAATTAAGTTCTGACTTGCTCACTCTATCCTGGATAGGCACTTGGGAACT TASACTCTTAAGCCATTCCAGTCATGATGAGGTGGAATGTATCAGTATACCA ATTAATATTTTTGAAAGAGCTCTTTTAGGTTAATTTAAGTacagcaatttctcatgtaatgttt a (SEQ ID NO: 584)

For: 5'-3' = ttagacaacttactactttgatgtcct (SEQ ID NO: 585) Rev: 5'-3'= taaacattacatgagaaattgctgt (SEQ ID NO: 586)

For: 5'-3' = teagacagtttagttggttacttce (SEQ ID NO: 588) Rev: 5'-3'= aagacacatectcagctaatettt (SEQ ID NO: 589)

#### M198 = DBY STS 08a (444 bp) C to T at position 45

For: 5'-3' = tgaggtggaatgtatcagtatacc (SEQ ID NO: 591) Rev: 5'-3'= tgatttcaaggatttgttagtctt (SEQ ID NO: 592)

M199 = DBY STS 08b (444 bp nominal) + 1 bp insertion (extra G) at position 404 (445 bp with mutation).

For: 5'-3' = tgaggtggaatgtatcagtatacc (SEQ ID NO: 594) Rev: 5'-3'= tgatttcaaggatttgttagtctt (SEQ ID NO: 595)

#### M200 = DBY STS 09a (429 bp) G to A at position 318

GgettacaettgcagactttgCAAATCTTAAGACTAACAAATCCTTGAAATCACACAGCTT GCAAATACGTACTAAACTGCACAAGGTGTGTGTTCTATATGTGCAGTTTTAGC GTATTTTAGTTGCATAGGTTTCCATGGTATTTATAGTCTCTTGTGCTAAATTTG GCCAAAGATGATTGTCCACCACTAAAAATGCCTCTCCCACTTGGAATTCTGTA CTGATTTTGTGGCCAGATGCAATGATCTTTAAAAACAAATCTTTTCAATGGCA TAAGAAGTTGACAAAAATTTCTTAAAGTGCAATAGATTTTCAARTTGT GCCTTGTTCTAAAACTTTTAAGTAGGTGCACTTGACAGTATTGAGGTCATTTG TTAAGGTGCTATTTCAATTAGTAGGTGCACTTGACAGTATTGAGGTCATTTG TTAAGGTGCTATTTCAATTAGTAGTGTAGGTTGTACAGTTTTCAATTAGTAGGTGCACTTGTACAGTTTTCAGTTTGT

For: 5'-3' = ggcttacacttgcagactttg (SEQ ID NO: 597)

Rev: 5'-3'= ggagaaatgtacaagagtctaaacc (SEQ ID NO: 598)

#### M201 (326 bp) DBY exon 11&12 G to T at position 136

 $TatgcatttgttgagtatatgtcAAATTGTGACACTGCAATAGTTACTACTTGAGTTACTATA\\TTAGTGCAATTAATTACACAACTATATATAGTAAttagtttctcagatctaataatccagTATC\\AACTGAGG{\color{red}K}TTTTCGTAATAGGTACTTAGTGTTGGATGAAGCTGATAGGATG\\$ 

CTGGATATGGGATTTGAACCTCAGATACGTCGTATAGTTGAACAAGATACTA TGCCACCAAAGGGCGTTCGTCACACCATGATGTTTAGTGCTACTTTTCCTAAG GAAATACAGGTACTGTTTGAcgtttgaactttcattcagaac (SEQ ID NO: 599)

For: 5'-3' = ttagtttctcagatctaataatccagt (SEQ ID NO: 600)

Rev: 5'-3'= gttctgaatgaaagttcaaacg (SEQ ID NO: 601)

M202 = DBY exon 16 (392 bp) T to G at position 259. Non-coding (cDNA bp# 1974+38)

For: 5'-3' = ggaattgcagggtttaagc (SEQ ID NO: 603) Rev: 5'-3'= gccacccacctgtaatcc (SEQ ID NO: 604)

M203 = UTY1 exon01 (1014) (503 bp) G to C at position 248; synonymous substitution, SER

GagtgccaagctgaggatgaCCCCGTCATCAACGTGGGCAAGCTGCGTCCAGGCCTTCC
CGGAGAGTATCGCCAGCCAACCAGGCGGGTGATGGAGGTGCGTACCTGTCCA
TGCCACCAAGCGCCTCCCTTTCCTCGACTGTCAGGCTAACAGACTCCTCTTCA
CTCTCGCGGCTCGCTTTTCCTTCCGCCATTTTCTTTGCCTCATCACCGAAGGCA
ACAGCGGCGGTAGTGAGCGACACTGCGCASGATTTCATGGAAACAACAAATT
TCCAAGTCCCACGACGATACCCAACCTTAATCGAGTAGTTGAAAAGACGCCT
TCAATCGCTGCTTGAGACTGTGACGCCAATTTTATCGCCTCCTCAGCGGCTGC
AAGGAAAAAAGCTGAGGCAAAGACTTAAGCTACCGAAGCACGGCAGCGGA
ACTCGGCTACCTGGATCACATCTGGGAAACTACAGGGAAGGCAGAAGCTCGC
AGTGCtggagagcacagcagaattt (SEQ ID NO: 605)

For: 5'-3' = gagtgccaagctgaggatga (SEQ ID NO: 606)

Rev 5'-3': aaattctgctgtgctctcca (SEQ ID NO: 607)

New Rev 5'-3':tccttggcagccgctgaggag (SEQ ID NO: 608)

M204 = UTY1 ex 02 = Intron 1 (1158-4) (286 bp) T to G at position 234; non coding AaggggcgaagtattccagAGTACGGGGACAGCAAAGGCAAAGACACTTTTCCGACC CCTTGGCCATGGAGCAGAGCCAAAATAAATACTGGCTGGGCGGTAAGGAAC GCGGGGCCTTGGTAGAGCAAAGTGCGGACCAAAGACTTTGCGTCTGCTTTTACCTTGCCTAGTAGGGTCTTCGTTCTGGCGCCATCTTCATGAAGCCTCAC GAACCCGAAGAGACGGCTGKAGAGAGAGAGAGACACAGAGCTTGTTAATGGTC TGAGAAAGCCAGTGACTTGCTCCTTCCCGAGTCCAAGAGCGACAGA TTGGTGAGTGCCAAGCTGAGGATGACCCCGTCATCAACGTGGGCAAGCTGCG TCCAGGCCTTCCCGGAGAGTATCGCCAGCCAACCAGGCGGTGATGGAGTG CGTACCTGTCCATGCCACCAAGCGCCTCCCTTTCCTCGACTGTCaggctaacagactcct cttca (SEQ ID NO: 609)

For: 5'-3' = aaggggcgaagtattccag (SEQ ID NO: 607) Rev 5'-3': tgaagaggagtctgttagcctg (SEQ ID NO: 608)

For: 5'-3' = gtataatactgtggttggaaagca (SEQ ID NO: 610) Rev 5'-3': ccaaactatgtgataataaatggg (SEQ ID NO: 611)

For: 5'-3' = gtataatactgtggttggaaagca (SEQ ID NO: 613) Rev 5'-3': ccaaactatgtgataataaatggg (SEQ ID NO: 614)

For: 5'-3' = aggaaaaatcagaagtatccctg (SEQ ID NO: 616)

Rev 5'-3': caaaattcaccaagaatccttg (SEQ ID NO: 617)

M208 = UTY1 = Intron 3b (1330+5798) (507 bp) C to T at position 352.

AtaaatacaaaatcacctgatggatATGCAAAAATTTATCAGCTTTACAAAGACATATAATA CCATTCTATGAGCACAAGTTTATTGCAATATTTTGTCCTTTACTGTCAACAAA AGAACACAGCCACATGATATAGGAAAAATCTATATTCTTTACAAATTTTCCAT GAATCTCTAGCTAAAAGATCATATGACATATATGCAACGATTTATCAGCTTTC AGAGCTTTAATTGATATTCATTACTTGTGGGTTCTGTTATTTGACTCACGAAA ATTTATATATACACAAAATCAATACTTAATGATGGTTTCAAAGATATTCACAG ACCTGCTCAGGGCAGCAATAAATTYGACCCACTGGATACACACACTCCCAGCTA ATGTTAGAAGCGGTGGGCCTTTCTCTGACTTCATGTGTCAAGTATTCTAAACA AACAGGCTTTTCCTGCTGTATGCAGTGTCACATTTTTCTGATTTTTGCTCttttgtta gtaatttcgctgtttaa (SEQ ID NO: 618)

For: 5'-3' = ataaatacaaaatcacctgatggat (SEQ ID NO: 619) Rev 5'-3': ttaaacagcgaaattactaacaaaa (SEQ ID NO: 620)

M209 = UTY1 = Intron 3c (1330+6211) (550 bp) A to G at position 471.

CactgtcttccacaatggttgAACTAGTTTACAGTTCCACCAACAGTGTATAAGTTTTCCT
ATTTCTCCATATCCTCTCCAGCACCTGTTGACATTACTAAAATAACATTCTCAT
CAAGGTCATCAGGGTCTCAGAACTGGCTACATACAACCTCCAAGAAAGTTTC
GTTCTTTCTGTTTTTGCAATGTGTTCTGCCACAAATTCATCAGTTCTCAAAGCT
AACAGAACTTTACTAGTTGCCCAATGCATCAATTCCATAGTTCTGAGAGCAT
GGGCATGAATGTCTGAAAACCTGAGGTATGATCACTAATATGCTATTCTCTGA
ACTTCTCAATTGCATTTTCCTCCTTGAATAAATCAGACTAAATTAGTGACACC
ACAAATTGTGATCATTGAGAAATCTCTAAAGGTTTTTCAGAAGCCGAGTAGG
AAGCTATCTATGACTTTTTAAAACTCTGACTGAATTCTRAATATTTAATTG
GACATTACATGAAGACGTTGTGTATTTAACTTCTGAATGCAgggaagataaatacaaaat
cacct (SEQ ID NO: 621)

For: 5'-3' = cactgtcttccacaatggttg (SEQ ID NO: 622) Rev 5'-3': aggtgattttgtatttatcttccc (SEO ID NO: 623)

M210 = UTY1 = Intron 3d (1330+6221) (550 bp) A to T at position 461.

CactgtettccacaatggttgAACTAGTTTACAGTTCCACCAACAGTGTATAAGTTTTCCT
ATTTCTCCATATCCTCTCCAGCACCTGTTGACATTACTAAAATAACATTCTCAT
CAAGGTCATCAGGGTCTCAGAACTGGCTACATACAACCTCCAAGAAAGTTTC
GTTCTTTCTGTTTTTGCAATGTGTTCTGCCACAAATTCATCAGTTCTCAAAGCT
AACAGAACTTTTACTAGTTGCCCAATGCATCAATTCCATAGTTCTGAGAGCAT
GGGCATGAATGTCTGAAAACCTGAGGTATGATCACTAATATGCTATTCTCTGA
ACTTCTCAATTGCATTTTCCTCCTTGAATAAATCAGACTAAATTAGTGACACC
ACAAATTGTGATCATTGAGAAATCTCTAAAGGTTTTTCAGAAGCCGAGTAGG
AAGCTATCTATGACTTTTTAAAACTCTGWCTGAATTCTAAATATTTAATTG
GACATTACATGAAGACGTTGTGTATTTAACTTCTGAATGCAgggaagataaatacaaaat
cacct (SEQ ID NO: 624)

For: 5'-3' = cactgtcttccacaatggttg (SEQ ID NO: 625) Rev 5'-3': aggtgattttgtatttatcttccc (SEQ ID NO: 626)

M211 = UTY1 = Intron 4a (1381+16283) C to T at position 381.

CaattcactatttgaggaatccaAGTATTCCCCCTGGGGCACAGTTTAGGTATAAACACACT TCCACTACTAACTATCTCCAGCAGTTGCCTACCTATAAGCTCCACCTACAGGC

For: 5'-3' = caattcactatttgaggaatcca (SEQ ID NO: 628)

Rev 5'-3': gaagtetetgatttatttggcag (SEQ ID NO: 629)

For: 5'-3' = tataatcaagttaccaattactggc (SEQ ID NO: 631)

Rev 5'-3': ttttgtaacattgaatggcaaa (SEQ ID NO: 632)

M213 = UTY1 ex05b = Intron 4c (1381-78) T to C at position 290. Mimics M89 (409 bp); non coding

For: 5'-3' = tataatcaagttaccaattactggc (SEQ ID NO: 634)

Rev 5'-3': ttttgtaacattgaatggcaaa (SEQ ID NO: 635)

CTGGTTACTTTCGTTCGTTTATTTTTCYGTTCTCAGACAGTGTCTCACACTGTCTCCCAGGctggagtgaagtggcatttc (SEQ ID NO: 636)

For: 5'-3' = tattacaaaatatggaaacaaggc (SEQ ID NO: 637)

Rev 5'-3': gaaatgccacttcactccag (SEQ ID NO: 638)

M215 = UTY1 exon 14 (2358) (386 bp) A to G at position 163; silent substitution, SER GtaaaactcagatatacacccatgAAATATACACAGAAACTATAAATTAGCATTAATATC CTCTAAAATGATACTGTAGTAAAGAAAATATTCTCAAACTGTTGGTAAATTTTA GAGAAAATAAAAAATATTATACATACTTGCTGCATTAAGACAAACTGRCTTTC TAACTGTTCCAGCTGATGCTTCTGTGCTGGATTTAAATTATCTCTATTTGCTCG CAGTTGTTCCAAGTGCTAGAAGAAAAAGAGATTAATAATCAAAAGTTTAATC TAAAATTTAAGACAATATAAGGCAACTCCTCACTAAAAAGACTACACAGAAC CTTTGCAGGATGAAAGACAGTGATTCCTAATGAACgttaagatagtgattcttttttttt (SEQ ID NO: 639)

For: 5'-3' = gtaaaactcagatatatacatcccatg (SEQ ID NO: 640)

Rev 5'-3': aaaaaaaaagaatcactatcttaacg (SEQ ID NO: 641)

**M216** = UTY1 intron 18 3678+537 (557 bp) **C** to **T** at position 54.

For: 5'-3' = ctcaaccagtttttatgaagctag (SEQ ID NO: 643)

Rev 5'-3': gagagtctgaactaatgtgtcttgt (SEQ ID NO: 644)

 $M217 = UTY1 \text{ intron } 17\ 3678+768\ (461\ bp)\ A \text{ to } C \text{ at position } 219.$ 

GcttatttttagtctcttccatGACTCTTCTAATACCATCGTCAATAAATTTCAACTAGGTA
AAAAATTAATTGAACATCTGTCCAAAGAAAGGCCAGTATCTCCAAAATCC
TCTCGTACAGATCTGTTTCGAGATCATTCTAATTACTGTATCTTCATATTTTAG
GTTAAGATTCTTTAACTTGTGAAGGAGAATGAAAAAGTTGGGTGACACMAA
CTCTTCAGAAGGAAAAATACATAAAAATTATTTTGATGAAAGCCACAGCAGC
TTTATCAAATGCTTACGTTGCTAAATAGTAAAAAAAGCCACTTAAATTCCAAT
GGAAATTTTATACCCACATGTATTTATGTAAAACTTTTAAATAACATGTATTC
ATAATCACTTTTATATCCTCAACCAGTTTTTATGAAGCTAGAAAAAAATTCCT
TTATTaaagaaatgtaacattcaacaggt (SEQ ID NO: 645)

For: 5'-3' = gettatttttagtetetetteeat (SEQ ID NO: 646)

Rev:5'-3': acctgttgaatgttacatttcttt (SEQ ID NO: 647)

 $M218 = UTY1 \text{ intron } 16\ 3679-281+768 (482 \text{ bp}) C \text{ toT} \text{ at postion } 380.$ 

For: 5'-3' = ttgtgagtttttttccatcaatc (SEQ ID NO: 649)

Rev 5'-3': tttattgacgatggtattagaagag (SEQ ID NO: 650)

 $M219 = UTY1 \text{ intron } 16\ 3676-294\ (482\ bp)\ T \text{ to } C \text{ at postion } 232.$ 

TtgtgagtttttttccatcatcTGGCTATTAAAAATCTGCAGTGCATCCTAACCTTTGATAT TATGTTGCTACATATTACAGTATTGTATCATTTGTCTTGTCAGGAAAGTGTGG AGGTAATAGCTAAAAAAAACCCTCTCTTTTAAAAATTACATTTTAAATTTGAT TCACTTTAAAACTGTTACCTATCTCTTATACCACAGTGATTTATAAAATTCTTT TAAATTAGYTGAGTTGTTCGAAAGTATTTCCCAAGCATATTTTTTGAGTTATC TTCTATTGCTTCTTAAATGAGACAACAGGTAGAAGAGACATTTAAAAGTTTAA AATCAAACTGTTTATAAAACTATTAACAAAACTTTTAGGGAATAAAAAACCAC AACAGGCAAACCTTAAATTTGTATTTATTGCCTCAAAGTTTCAACTGAAACGC TTATTTTAGTCTCTCTTCCATGACtcttctaataccatcgtcaataaa (SEQ ID NO: 651)

For: 5'-3' = ttgtgagtttttttccatcaatc (SEQ ID NO: 652)

Rev 5'-3': tttattgacgatggtattagaagag (SEQ ID NO: 653)

 $M220 = UTY1 \text{ intron } 16\ 3676-329 \ (482 \text{ bp}) A \text{ to } G \text{ at postion } 367.$ 

TtgtgagtttttttccatcatcTGGCTATTAAAAATCTGCAGTGCATCCTAACCTTTGATAT TATGTTGCTACATATTACAGTATTGTATCATTTGTCTTGTCAGGAAAGTGTGG AGGTAATAGCTAAAAAAAAACCCTCTCTTTTAAAAATTACATTTTAAATTTGAT TCACTTTAAAACTGTTACCTATCTCTTATACCACAGTGATTTATAAAATTCTTT TAAATTAGCTGAGTTGTTCGAAAGTATTTCCCAAGCATATTTTTTGAGTTATC TTCTATTGCTTCTTAAAATGAGACAACAGGTAGAAGAGACATTTAAAAGTTTAA AATCAAACTGTTTTATAAACTATTAACAAAACTTTTAGRGAATAAAAACCAC AACAGGCAAACCTTAAATTTGTATTTATTGCCTCAAAGTTTCAACTGAAACGC TTATTTTAGTCTCTCTTCCATGActcttctaataccatcgtcaataaa (SEQ ID NO: 654)

For: 5'-3' = ttgtgagtttttttccatcaatc (SEQ ID NO: 655)

Rev 5'-3': tttattgacgatggtattagaagag (SEQ ID NO: 656)

M221 = UTY1 intron 18 (3784+165) (324 bp) G to A at position 200.

For: 5'-3' = gggaaatgtgaaaggaaaata (SEQ ID NO: 658) Rev 5'-3': ttaaacttttataacactgacagaaac (SEQ ID NO: 659)

M223 = A8.05e (208 bp) C to T at position 67.

For: 5'-3' = ttcagcaagagtaagcaagagg (SEQ ID NO: 661)

Rev 5'-3' = cctttttggatcatggttctt (SEQ ID NO: 662)

M224 = B9.60b (301 bp) T to C at position 193

CttcaggcattattttttttggtTCTCCACTACAGGAGAAATGTAAATGTGATGAGTCAGAAT TTAGGATGGCTGTATGGGTTTCTTTGACTAATACAAGAAATCACTTTGTAATG AATGAAATCAGTGGTTTCTGCATTACTCCGTATGTTCGACATGAACACAAATT GATACACTTAACAAAGATACTTCTTTCYGCCCTTCCAAATATTTCAAAATAAG CTGGTCATAGTACTTGCTTTTCATAAAAAGATGGTAAGCTTCCAATATTTAGA TTTaaggaaaggtgaaggaacactat (SEQ ID NO: 663)

For: 5'-3' = cttcaggcattattttttttgg (SEQ ID NO: 664)

Rev 5'-3' = atagtgttccttcacctttcctt (SEQ ID NO: 665)

M225= UTY1 Exon1b, (528 bp) **G to A** at position 369. (518 C to T in cDNA utr region AaggaaaaaagetgaggcaAAGACTTAAGCTACCGAAGCACGGGCAGCGGAACTCGGC TACCTGGATCACATCTGGGAAACTACAGGGAAGGCAGAAGCTCGCAGTGCTG GAGAGCACAGCAGAAATTTCTTAAAATCACAAACTTTGCCAGCACCACCACAA AGTTGTAATTGTGTCACGGGCGAACCCCACGCAGCAGCCGCCGCGACCTCCCCGC TCCCAACCACTTAGTTGTAGCCAATCTAGGCGACTGATTCGTCTCACGTGATC TTTGTTGACTTACGTCAGGCATTGCTCCACTGTACTCCTAGGCTGCTGGGACC CCGCCCAGCCAGTTCGCCAAGGACCTAGGAACATGACAGAGGCTGACT**R**ATT CTGACCGCTGGTTGGTTGATGGTCACGTCTATGGAGAAAAGGGTAGTCTCTG GGATGGAACAACCTGTAGGTTGTTGTTGCTAGTTAAATGCATTAAGATAGAAAATG GAGTGTCTGTGCTGGGTTTTTTTGCAGTTGCGatacgcttgaaggggaagag (SÈQ ID NO: 666)

For 5'-3'= aaggaaaaaagctgaggca (SEQ ID NO: 667)

Rev 5'-3'= ctcttccccttcaagcgtat (SEQ ID NO: 668)

M226 UTY Ex1c 1104 silent/glu (380 bp) C to T at position 158

For 5'-3'=gagtgccaagctgaggatg (SEQ ID NO: 670)

#### Rev 5'-3'=aaattctgctgtgctctcca (SEQ ID NO: 671)

M227 UTY Ex1c 1105 Glu/Gln C to G in at position 157

For 5'-3'=gagtgccaagctgaggatg (SEQ ID NO: 673)

Rev 5'-3'=aaattctgctgtgctctcca (SEQ ID NO: 674)

M228 UTY Ex1c (380 bp) 1106 Glu/Gly T to C at position 156

For 5'-3'=gagtgccaagctgaggatg (SEQ ID NO: 676)

Rev 5'-3'=aaattctgctgtgctctcca (SEQ ID NO: 677)

M229= UTY1 Int12, A to C at position 159. (1560+7060 T to G in intron6)

Group I

For 5'-3'=ggtacacacctgtagtcccaac (SEQ ID NO: 679)

Rev 5'-3'=cattttttagaactctttcatggtaa (SEQ ID NO: 680)

M230 (449 bp) UTY Ex9 intron 8 1651-143 T to A at position 367

Group VIII

AatgtcacatttagtcttaacccatAGACTTCTAAATGAAAACAAATGTCTAAGCAGAGGGA AAAAAATTGAACCTCAAAAGGCAAATCTCTTCAAATTAATGTAATGTAATAAT

For 5'-3'=aatgtcacatttagtcttaacccat (SEQ ID NO: 682)

Rev 5'-3'=acattattagtatgtaaatcttcattgc (SEQ ID NO: 683)

# M231 UTY Ex13 Intron 13 2283+33 G to A at position 110 in Group VIII

CctattatcctggaaaatgtggGCTCGTTTTAATTATTCATATTAATTTAGTTAATCATC ATTCAATTAATACCTAAAAAAACAACATTTACTGTTTCTACTGCTTTC**R**AATTG GGGGAAAGATCGTCAAAGAATTCATACCTGTAATTTCTGTGGTGTCAAACAC AACGAATAAACTTGCTGTACTGGATGATGTGAAAGACTCTGGCCACCATTCC AGTTATCAGAACCATTCTAAGGAAAAATTTAGTGTAAAAGATTAAGAATATTT GCTTAATTTCATACACTTAGAGTTATGACTAGTGAGAACCaagtgactaggaat (SEQ ID NO: 684)

For 5'-3'= cctattatcctggaaaatgtgg (SEQ ID NO: 685)

Rev 5'-3'=attccgattcctagtcacttgg (SEQ ID NO: 686)

# **M232** = UTY1 intron 17 3679-566 (461 bp) **C** to **T** at position 38 Group VIII

gettatttttagtctctcttccatGACTCTTCTAATAYCATCGTCAATAAATTTCAACTAGGTA
AAAAATTAATTGAACATCTGTCCAAAGAAAGGCCAGTATCTCCAAAATCC
TCTCGTACAGATCTGTTTCGAGATCATTCTAATTACTGTATCTTCATATTTTAG
GTTAAGATTCTTTAACTTGTGAAGGAGAATGAAAAAGTTGGGTGACACAAAC
TCTTCAGAAGGAAAAAATACATAAAAAATTATTTTGATGAAAGCCACAGCAGCT
TTATCAAATGCTTACGTTGCTAAATAGTAAAAAAAGCCACTTAAATTCCAATG
GAAATTTTATACCCACATGTATTTATGTAAAACTTTTAAATAACATGTATTCA
TAATCACTTTTATATCCTCAACCAGTTTTTATGAAGCTAGAAAAAAATTCCTT
TATTaaagaaatgtaacattcaacaggt (SEQ ID NO: 687)

For 5'-3'=gcttatttttagtctctcttccat (SEQ ID NO: 688)

Rev 5'-3'= acctgttgaatgttacatttcttt (SEQ ID NO: 689)

# M233 = UTY1 Exon18n, T to C at position150, (3784+37 A to G at intron18) Group III

AtcacttgcatcagtgctaaagaTGCTTGCTCATGCACAAGAGGTATAAAATTGAGTGAGA AAGAAAGATAACACACATTAAAATAAAGACTCAGAATGTTGGGGGAAAAAAT CAGTGAGTTTCTGTCAGTGTTATAAAAGTTTAAAGAYAGTAAAAATATATTC AATCTTGGTTTTAAGCTTACCTAATTTAAGAGCTCCAGCAAGGCCACGTATTA CTGTAACAGGGTTTTTTGGATttgtacaaaattgatgtaatggagGAAAGAAAGCATCACGTT TATTTTCCAACTGTAAAAGCAAAATATTTTGTTAGGTCTCAGATAAATGACAA AATATACCTCAGATTTGTGCCTTTAATAAAATGATTAAATACAATACTTCAAA

TTTGTGAGTTTTTTCCATCAATCTGGCTATTAAAAATCTGCAGTGCATCCtaacct ttgatattatgttgctacat (SEQ ID NO: 690)

For 5'-3'=atcacttgcatcagtgctaaaga (SEQ ID NO: 691)

Rev 5'-3'=atgtagcaacataatatcaaaggtta (SEQ ID NO: 692)

**M234=** UTY1 Exon20n, **C** to **T** at position 253, (4049 G to A in cDNA, codon 1015, Arg/Gln)

Group III

For 5'-3'=tctccattagcaatgtgtgtttt (SEQ ID NO: 694)

Rev 5'-3'=caataccaggcttcctcctt (SEQ ID NO: 695)

M235 = (317 bp) DFFRY Exon4, T to G at position 155. (1859 in cDNA, codon 65, Asp to Glu

For 5'-3'= tagatatttttccttaatctgtggt (SEQ ID NO: 697)

Rev 5'-3'= actacaatctactcttttctttttctc (SEQ ID NO: 698)

M237= DFFRY Exon30, (366 bp) G to C at position 39. (5903-132 in intron29)

Group III, 325 bp w/out homopolymer region in STS.

For 5'-3'=ttgcatttactgttctagagagttct (SEQ ID NO: 700)

newRev 5'-3'=gtacttactttaatgagattagcac (SEQ ID NO: 701)Homopolymer clipped off

M238= DFFRY Exon43, C to G at position 28 (8729-54 in intron42)

#### Group I

For 5'-3'=gtactaaatggcacataattaggaa (SEQ ID NO: 703)

Rev 5'-3'= aatttagaatgattttcagtatcagc (SEQ ID NO: 704)

M239 = DFFRY Exon43, G to A at position 148 (8795 in cDNA, codon 2377, silent/Ser Group I

GtactaaatggcacataattaggaaCTSAAATGTTAGCTACTATTGGATATTACAAAGTTTTACATCACTTCTGTTTTAGAATTCATAATGCACTATTGGATACTAAAGGAATTCCAGATGACAGAGATGGGCTGTTCGATACAATACAGCGCTCRAAGAATCACTATCAAAAACGAGCATATCAGTGCATAAAAATGTATGGTAGCTCTATTTAGCAGTTGTCCTGTTGCTTACCAGATCTTACAGGTGAGGGTTTTTCTCTTATAAATTTGTAGAAACCTCTGTCACAAGTAAGGAAATGATCGTGAAATTTTTGTATTAGCATTTTAAgctgatactgaaaatcattctaaatt(SEQ ID NO: 705)

For 5'-3'=gtactaaatggcacataattaggaa (SEQ ID NO: 706)

Rev 5'-3'= aatttagaatgattttcagtatcagc (SEQ ID NO: 707)

#### M240 = DBY int2n, C to T at position 47, (116+613 in intron1.)

CtgtggaattcttgaagacgagTGACTATAATATAGCACAACGTAAYAAGTATCCTGTATC TTGTTTCTGGTGGGGTCCCGTAGCCACGGAGCAACCGTTGCCCGGGTGCTGAG CGTGCCGAAACTGGGCTTCCGGTATGGAAAGTTTTGTGACGCAGAAGGACCG GAAAGGGATGGTGGGGAGGGTAGGGAAGGATGGCTGCCGCGTGCTTCTCTTG ACCCTGTAGAAATAATGGAAATTGGACGCCCGCGGAAAGACACCCTGGAAGGT TAGAGATCCAGCATTGCGCTACACCCCTTTGTTAATTCAGTCACTGGACAGCC GCCTAGCCGAGAGGTTTTTATATGGTATTGTATCTTTACTTTAGGCG ATACATGCAGAAGTCGTCCGGTAgaaaactaacctcgaatgttgatt (SEQ ID NO: 708)

For 5'-3'=ctgtggaattcttgaagacgag (SEQ ID NO: 709)

Rev 5'-3'=aatcaacattcgaggttagttttc (SEQ ID NO: 710)

### M241 DBY Intron 4 (intron 1) G to A at position 57 cDNA# 117-989

5'-3' For aactettgataaaccgtgctg (SEQ ID NO: 712)

5'-3' Rev tccaatctcaattcatgcctc (SEQ ID NO: 713)

M242 DBY Intron 4 (intron 1) C to T at position 337 cDNA# 117-866 Group X

5'-3' For aactettgataaaccgtgctg (SEQ ID NO: 715)

5'-3' Rev tccaatctcaattcatgcctc (SEQ ID NO: 716)

### **M243**= DBY int6, (401 bp) **T to C** at position 142, (117-356 in intron1)

Group III

For 5'-3'=ttttgagcttttgatgtttagga (SEQ ID NO: 718)

Rev 5'-3'=caaacttagctgccatgtcg (SEQ ID NO: 719)

## **M244**= DBY int6, (401 bp) **A to C** at position 174, (117-323 in intron1)

Group I

For 5'-3'=ttttgagcttttgatgtttagga (SEQ ID NO: 721)

Rev 5'-3'=caaacttagctgccatgtcg (SEQ ID NO: 722)

#### **M245**= DBY int8, **del AAACA** at position 264, (174+779 in intron2)

Group I

ATATTTAAATGGTAGAGACATTTTTGGATATTTAGTTAAATCCAAAAGTAGGAGGTTTAGTTCAAATTTGGATTTTTGAGTTAcaaaatcaggtagttaagtactgtcta (SEQ ID NO: 723)

For 5'-3'=gacgaagaacctaacattcagtg (SEQ ID NO: 724)

Rev 5'-3'=tagacagtacttaactacctgattttg (SEQ ID NO: 725)

### **M246=** DBY int8, **T to G** at position 284, (174+799 in intron2)

Group I

For 5'-3'=gacgaagaacctaacattcagtg (SEQ ID NO: 727)

Rev 5'-3'=tagacagtacttaactacctgattttg (SEQ ID NO: 728)

**M247**= DBY int9n, **T to C** at position 224, (175-693 in intron2)

Group II

For 5'-3'= atggtagagacatttttggatattt (SEQ ID NO: 730)

Rev 5'-3'=gcctgtaatcccagcacttt (SEQ ID NO: 731)

#### **M248=** DBY int9n, **T to C** at position 494, (175-444 in intron2)

Group VI

For 5'-3'= atggtagagacatttttggatattt (SEQ ID NO: 733) Rev 5'-3'=gcctgtaatcccagcacttt (SEQ ID NO: 734)

**M249**= DBY int10, **A to G** at position 313, (175-167 in intron2) Group II

For 5'-3'=tttcaccttgttagccaggat (SEQ ID NO: 736)

Rev 5'-3'=caagatactgcttttagaaaatgtct (SEQ ID NO: 737)

**M250**= DBY intl1n, **A to G** at position 299, (223+687 in intron3)

Group III

For 5'-3'=taacagttgttaagattaccactttt (SEQ ID NO: 739)

Rev 5'-3'=aacctttgatgcaattccag (SEQ ID NO: 740)

For 5'-3'=aaatattgcatctggctgga (SEQ ID NO: 742)

Rev 5'-3'=aagtccaaaagtaactatgtaatctt (SEO ID NO: 743)

New Rev 5'-3'=aatgacaagagtaaactcac (SEQ ID NO: 744) to exclude poly T region

M252=DBY int12n, (419 bp)ins T at position 354, (223+1127 in intron3. (site b) Homopolymer 7T's to 8T's

Group VI.

For 5'-3'=aaatattgcatctggctgga (SEQ ID NO: 746)

Rev 5'-3'=aagtccaaaagtaactatgtaatctt (SEQ ID NO: 747)

M253 = DBY int13, (400 bp nominal) C to T at position 283

Group VI

For 5'-3'=gcaacaatgagggtttttttg (SEQ ID NO: 749)

Rev 5'-3'=cagetceacetetatgeagttt (SEQ ID NO: 750)

M254 = DBY int13, (400 bp nominal, 418 bp derived)18bp INSERTION + 2bp substitution, A to G and G to C at positions 339, 340

Group VIII

For 5'-3'=gcaacaatgagggtttttttg (SEQ ID NO: 752)

Rev 5'-3'=cagetceacetetatgeagttt (SEQ ID NO: 753)

M255= DBY int14, (within derived 471 bp) C to T at position 107, (224-813, in intron3) Group V

TGACCTTGTGATCTGCCTGCCTTAGCC CTCCCAAAGTGCTGGGATTACAGGT GTGAGCCATCCCTGTTTTAATCCATCTGACATATTTCTTCTGATTATGTAGCTC TCTTAGTTCAAGCTTTCTGTAGGTAACCCACAGTCCCTGAGGTAATCTTTTA CTTAGCTGGGCCTTCCCAAAATGTGTATTATATATATAGCATATGTTAAATGTTT AGGTTTAACACCTtttgtattattcaggatttgtcaag (SEQ ID NO: 754)

For 5'-3'=ttttttttgagacggagtcttg (SEQ ID NO: 755)

Rev 5'-3'=cttgacaaatcctgaataatacaaa (SEQ ID NO: 756)

M256 = DBY int14, (derived 471 bp) ins  $\mathbb{C}$  at position 249, (224-672 in intron3) Group V

ttttttttgagacggagtettgCTGTGTTGTCCAGGCTGGAGTACAGTGGCGCGATCTCAGC
TCACTGCAAGCTCCACCTCTTGGGTTCATGCCATTCTCCTGCCTCAGGCTCCT
GAGTAGCTGGGACTACATAGGTGCCCGCCACCATGCCCAGCTAATTTTTTTGT
ATTTTTAGTAGAGACGGGGTTTCACCGTGTTAGCCAGGATGGTCTTGATCTCC
TGACCTTGTGATCTGCCTGCCTTAGCCCTCCAAAGTGCTGGGATTACAGGT
GTGAGCCATCCCTGTTTTTAATCCATCTGACATATTTCTTCTGATTATGTAGCTC

TCTTAGTTCAAGCTTTCTGTAGGTAACCCACAGTCCCTGAGGTAATCTTTTA CTTAGCTGGGCCTTCCCAAAATGTGTATTATATATAGCATATGTTAAATGTTT AGGTTTAACACCTtttgtattattcaggatttgtcaag (SEQ ID NO: 757)

For 5'-3'=ttttttttgagacggagtcttg (SEQ ID NO: 758)

Rev 5'-3'=cttgacaaatcctgaataatacaaa (SEQ ID NO: 759)

**M257**= DBY int14, (nominal 470 bp) **T to C** at position 373, (224-547 in intron3) Group I

For 5'-3'=ttttttttgagacggagtcttg (SEQ ID NO: 761)

Rev 5'-3'=cttgacaaatcctgaataatacaaa (SEQ ID NO: 762)

**M258**=DBY int15, (475 bp) **T to C**, at position 123, (224-388, in intron3) Group VI

GGACTTGTAGACCATTGGTCCCATAAAGATAACATAAAGGAAGACTACATGT GAAGGACTTCATATTTTgaaagatgcaaattattcaaaagtc (SEQ ID NO: 763)

For 5'-3'=tatatagcatatgttaaatgtttaggt (SEQ ID NO: 764)

Rev 5'-3'=gacttttgaataatttgcatctttc (SEQ ID NO: 765)

**M259**= DBY int16, (396 bp)**T to G** at position 151, (352+271, in intron4 Group IX

CagaatgttggtttactcattgttTTGTTAGCAGTAAGAGGTCTTTATTAATTTATTAAATTA GATGAATATGGTATTTGACACAGTGAAATCTGTTTCAACTTAAATGATACTTA AAGCCTGTCTGTGACAGCTTTAAACACTTCATTTKTGATGTGTGTTATAAGTT GATCTTAAAAACCTAATGGCTGTATTTAATCCTTTCTGTTTTTCACAAATAGG AGTAAAACTCTAAAAATATTCTCTTGTCACATGTCTACTTTCATATAAAGGAG AAATTCAAGTGTTATTCCTGCTTTCCTACTAGTAAATATTTAGATGATACT ATTTTAAATGAAGATGTAAAGTACGTAACTAGTTATAAGTATCTaaaaaccctaattctt agcatgtga (SEQ ID NO: 766)

For 5'-3'=cagaatgttggtttactcattgtt (SEQ ID NO: 767)

Rev 5'-3'=tcacatgctaagaattaggttttt (SEQ ID NO: 768)

**M260**= DBY int19, (343 bp) **G to A** at position 253, (608-124 in intron6) Group VI

CcacacccagctcatttttGTACTTTTAGTAGAGACAGGGTTTCGCCATGTTGGCCAGGCTGGTCTCAAATTCCTGATCTCAAGTGATCTTCATGCCTTAGCCTCCCAGAGTGCTGGGACTACAGGCATCAGCCACCATACCTGGCCTCCAAAAACTTTTTTCAATGTAGATTAAAACCCAGGCATTTTCTTAAAAAAATGCCATGAATCTTTTACTGAAATCATAGCATCTGTAAACTAAATCAGACAGTTTARTTTTTTTTTAGTAAATGTTAGTATAAAACAGAAATTGCGACAGATACAGCATTTTATATCtgctatgtttacttctgtatttactt (SEQ ID NO: 769)

For 5'-3'=ccacaccageteattttt (SEQ ID NO: 770)

Rev 5'-3'=aagtaaatacagaagtaaacatagcag (SEQ ID NO: 771)

**M261=** DBY int22, (284 bp) **A to G** at position 213, (1090-32 in intron10) Group X

For 5'-3'=atttgaggetetgagettea (SEQ ID NO: 773)

Rev 5'-3'=catatccagcatcctatcagc (SEQ ID NO: 774)

**M262=** DBY STS01, (502 bp) **del** A at position 226, (1-2908 out side of 5' region) Group III

agctgtttggacttgagttgTAGAATAACTGAAAATAGGAAACTGCTATATATATATGT ATGTATAATATATATACCTTTTTTCAGGTACTCCTATTGCAATACCTGCATTT CAGCACTATTCAAAAGTAAAATAAGTCCCAGAGCCAGGTTAGTCATTATGTC CTATTTATTGCTAATTTTCATATACAAATGAGAGCTGTCAGAATTCACAGCTT CTGAATATCAGAAGCTCATGTTTTCCCTGGTCTATACAAAAAGGAAATAAGT GAGGCCAAAAATGTACTTTAACAGTGCTCCATAATACGAATCTCATAAATGA GCTGGAATAGACCCTGAGGTCTTCAAGCCTAGTTTCTCAAGATCGTATTTTGT AAACTTGTGCTAGCAGTTTTGAATATCACAATGATTGGCATGGGCTGCTGACA TTTTAGCAGGCAGGCTCAGGGTGTTAGATGTCCTGTAATTCAGGgacattcacagta gaaaatactttgg (SEQ ID NO: 775)

For 5'-3'= agetgtttggaettgagtagttg (SEQ ID NO: 776)

Rev 5'-3'= ccaaagtattttctactgtgaatgtc (SEQ ID NO: 777)

M263=DBY STS06, (515 bp) G to C at position 332, (1-341 out side of 5' region) Group III

For 5'-3'= ccactcagetttcctcaggt (SEQ ID NO: 779)

Rev 5'-3'= cgttcgtttagtcataagatcg (SEQ ID NO: 780)

**M264**=DBY Exon17, (552 bp) **C to T** at position 115, (1988 at cDNA, codon639, silent/Gly)

Group III.

For 5'-3'=tccaactctagatttcttttactgg (SEQ ID NO: 782)

Rev 5'-3'=ggtatcgtgagcattactgagc (SEQ ID NO: 783)

M265= DBY STS07, C to A at position 298, (2312+358 outside 3' region) ttagacaacttactactttgatgtcctGTTGGCTCAGTAATGCTCACGATACCAATTGTTTTGAC AAAATAAATTTACTAAACTTGGCCTAAAATCAAACCTTGGCACAGAGGTATG ATACAACTTTAACAGGAGTCATCAATTCATCCATAAATATAAAAAGGGAAAA

AAACTTAAGGCAGTAGTCTGCATTAGGACTGTTTGAGTTTTGCAGACTTGGGG TTGGGAGAACATCTTAAAGCATTAAAGCATAGTTTTTTGTATGGCCAACCTTA CTAAATTAAGTTCTGACTTGCTMACTCTATCCTGGATAGGCACTTGGGAACTT ACACTCTTTAAGCCATTCCAGTCATGATGAGGTGGAATGTATCAGTATACCA ATTAATATTTTTGAAAGAGCTCTTTTAGGTTAATTTAAGTacagcaatttctcatgtaatgttt a (SEQ ID NO: 784)

For 5'-3'=ttagacaacttactactttgatgtcct (SEQ ID NO: 785)

Rev 5'-3'=taaacattacatgagaaattgctgt (SEQ ID NO: 786)

**M266**= DBY STS08, (444 bp) **T to C** at position 208, (2312+623 outside 3' region) Group II

For 5'-3'=tgaggtggaatgtatcagtatacc (SEQ ID NO: 788)

Rev 5'-3'=tgatttcaaggatttgttagtctt (SEQ ID NO: 789)

M267 EIF1A Y STS12 (site a) (287 bp) T to G at position 148. STS also contains two Group I associated mutations

newFor 5'-3=ttatcctgagccgttgtccctg (SEQ ID NO: 791)

Rev 5'-3'=tgtagagacacggttgtaccct (SEQ ID NO: 792)

**M268** = EIF1A\_Y STS5a, (427 bp) **A to G** at position 292, GROUP VII

ctaaagatcagagtatctccctttgCAAAATGTCCATTAAATCTTTGCTGATGTTATTATCCCT GTACCTGACTCTATCCTTAAATAGTAAGGCTTCCTTTATTCTTGTAGGGTAGA ACTTTTAAACTGAGTGATGCCTAAAAATGTTCTCAATAAAGAGAGTATCTCCA AAACACGTCGGATTTGTTTAAAGGGAAGTGTGGATTTTTTGATCTTAGAAA GGAAACGAGATAAAATATTAAACGACTTTAATTTTTGTATGATCATGCCTAGC CTCATTCCTCTAAAAT**R**TAATTTAAAGTGGATTCTGTTACATGGTATCACAAT AGAAGGGGAATGATCAGGGTTTGGTTAATTCTGGTAAATTGAAAACAATTTT TTTTTT(T)ATCATATGTGCCTCAgaaggcacacaaaagaagtatagt (SEQ ID NO: 793)

For: 5'-3'= ctaaagatcagagtatctccctttg (SEQ ID NO: 794)

Rev: 5'-3'= actatacttcttttgtgtgccttc (SEQ ID NO: 795)

**M269** = EIF1A\_Y STS5b, (427 bp) **T** to **C** at position 358, Group IX

CtaaagatcagagtatctccctttgCAAAATGTCCATTAAATCTTTGCTGATGTTATTATCCC TGTACCTGACTCTATCCTTAAATAGTAAGGCTTCCTTTATTCTTGTAGGGTAG AACTTTTAAACTGAGTGATGCCTAAAAAATGTTCTCAATAAAGAGAGTATCTCC AAAACACGTCGGATTTGTTTAAAGAGGAAGTGTGGATTTTTTGATCTTAGAA AGGAAACGAGATAAAATATTAAACGACTTTAATTTTTGTATGATCATGCCTA GCCTCATTCCTCTAAAATATAATTTAAAGTGGATTCTGTTACATGGTATCACA ATAGAAGGGGAATGATCAGGGTTTGGTTAAT YCTGGTAAATTGAAAACAATT TTTTTTT(T)ATCATATGTGCCTCAgaaggcacacaaaagaagtatagt (SEQ ID NO: 796)

For: 5'-3'= ctaaagatcagagtatctccctttg (SEQ ID NO: 797)

Rev: 5'-3'= actatacttcttttgtgtgccttc (SEQ ID NO: 798)

M270 = EIF1A\_Y STS5, (428 bp) ins T at position 387.. Has ancestral T at M281. HOMOPOLYMER

For: 5'-3'= ctaaagatcagagtatctccctttg (SEQ ID NO: 800)

Rev: 5'-3'= actatacttcttttgtgtgccttc (SEQ ID NO: 801)

M271 = UTY1 intron 17 3679-566 (461 bp) A to C at position 296
Group VIII. Discovered while typing M232. This STS also contains M217 site.
gcttatttttagtctctcttccatGACTCTTCTAATACCATCGTCAATAAATTTCAACTAGGTA
AAAAATTAATATTGAACATCTGTCCAAAGAAAGGCCAGTATCTCCAAAAATCC
TCTCGTACAGATCTGTTTCGAGATCATTCTAATTACTGTATCTTCATATTTTAG
GTTAAGATTCTTTAACTTGTGAAGGAGAATGAAAAAGTTGGGTGACACAAAC
TCTTCAGAAGGAAAAAATACATAAAAAATTATTTTGATGAAAGCCACAGAGCT
TTATCAAATGCTTACGTTGCTMAATATTTATGTAAAAAAAAAGCCACTTAAATTCCAAT
GGAAATTTTATACCCACATGTATTTATGTAAAAACTTTTAAAATAACATGTATTC
ATAATCACTTTTATATCCTCAACCAGTTTTTATGAAGCTAGAAAAAAATTCCT
TTATTaaagaaatgtaacattcaacaggt (SEQ ID NO: 802)
Rev :5'-3': acctgttgaatgttacatttcttt (SEQ ID NO: 803)

M272= EIF1A\_Y STS4, (496 bp) A to G at position 212, GROUP VIII

CaggagggaccatgttttATAGTCCACAAAAACTCTGTTTAGATTATTCCTTCCTGGGA CCCAGACCAATTTGTCTTCTTTTACTTGCCTGTTGGCAGCATGGAATCTGTTT CATTTCTCTTTTTAGCTGTCACGACACACACACTCTTGAGGTACTTGGTGACA

For 5'-3'= caggagggaccatgtttt (SEQ ID NO: 805)

Rev 5'-3'=cagcaaagatttaatggacattt (SEQ ID NO: 806)

#### M273= EIF1A STS8, (502 bp) C to G at position 189 GROUP II

CacatcaggaaaagggcatcCTTTGGCCTATACTTGTGAAGAGCTAGAGTAAGGTGCTC CCCACCTTTGAGATTGCTAAAGTTGTCATTCTTTTTGGAAATTTATGAGCTAAT CATCATTTAGTCATTTGAAAAGCTGCCAAACTTTTGTAAAACCCAGTAAGGA AAGCAGGTATGATCTTTGTCCTGASGCAGCTAAGTTCAGGCACGATTAATTGC TCGAAATATAGAATGTGTTTTCCTTTGTAGAAATTTAGTTTTGGCATGCCCTA AAATGCATCAGAATCTGGATAAATCACAGAGTTCTGGAAGCCCAATTGTCTT CTATAGTGGCACAGAACAATGTGAGACTGCCCCAGAGGTAGTGTGGAATTC AAGAAGTTAGATGTCTGGCTTTATGGTGGCCAGGTATATGTTTTATTCTATTT GCAGTGTTAACATTTTTATTCAAATTCTTCAATCGATCCCTTAATATTACTGTA atttgtagectttctccctcc (SEQ ID NO: 807)

For 5'-3'=cacatcaggaaaagggcatc (SEQ ID NO: 808)

Rev 5'-3'=ggaggagaaaggctacaaat (SEQ ID NO: 809)

#### M274= EIF1A\_Y STS2a, (457 bp) C to T at position 47, GROUPVIII w/M11

For 5'-3'=gccatgcccaagaataaag (SEQ ID NO: 811)

Rev 5'-3'=ctaaacatgcttcaaggcttc (SEQ ID NO: 812)

## **M275**= EIF1A\_Y STS2b, (457 bp) **C** to **G** at position 325 GROUP X

gccatgcccaagaataaagGTACTGCTGTAAGCCTCTGGGACTATAYCTCGGCTTGCTCTGCCAGTAACCCCGACGCCTGTTCCAGGCCGCAGTGACTGTTCTAACGGCGGTACTGGCCACTGCGACCCCAGCACTGTGTTCGGGAAAGGAGCTGGGAATGCCCTATTTGGTCACATTGGGGGTGGGACAGACGCCATTTTTGTGGGGCCTCCTTCGGAAGATAGCGGGCTTTTTGCTGCTGCTGATTTCACGCCAGACGGAAAACGTATAGGT

AGGGACGGTTGAGGGACCTTAACCGGACGGCCTGGCTTTCCAGAATAGGCAC ATGSAAACACTTCCCTGCTACTTTCCTGGAAGCGGTTCTTAACTTTGAAGACT TACCTATCTGGACAGTTAAAAGTATTGCTAAGGATACTCCCTTTTCCTTGTTA AACAGTGGGgaagcettgaagcatgtttag (SEQ ID NO: 813)

For 5'-3'=gccatgcccaagaataaag (SEQ ID NO: 814)

Rev 5'-3'=ctaaacatgcttcaaggcttc (SEQ ID NO: 815)

**M276** EIF1A\_Y STS12 (site b) (287 bp) **T** to **A** at position 58.

Group I associated mutation. Has another Group I site (M277) and a Group VI site (M267).

ttatcctgagccgttgtccctgTGTTTCCATTTCTCTTTTCCTCATTTCTCATCATC**W**ACATT TCTCCTGTACTTGTTCATTAAATAATGATTCCTTGGATATACCAAGTCTGGAT AGCGGATTCGATGGAAGCATTTTTGTAAATATACGTTCAGTATTTTGTGTGGA AGAACACAATCTAGCTGATGCCTGCAATCCCAGCCCTTTGGAAAGCGAGGTG GGTGGATTGCTTGAAGCTACGAGTTTGACACTAGCCTGGGCAACagggtacaaccgt gtctctaca (SEQ ID NO: 816)

newFor 5'-3'=ttatcctgagccgttgtccctg (SEQ ID NO: 817)

Rev 5'-3=tgtagagacacggttgtaccct (SEQ ID NO: 818)

M277 EIF1A\_Y STS12 (site c) (287 bp) G to T at position.

Group I associated mutation. G to T at position 151. Has another Group I site (M277) and a Group VI site (M267).

newFor 5'-3=ttatcctgagccgttgtccctg (SEQ ID NO: 820)

Rev 5'-3'=tgtagagacacggttgtaccct (SEQ ID NO: 821)

M278= DBY int12n, site c ((nominal, 418 bp)) **T** to **G** at position 374, Site within STS with 7 T homopolymer.

Group I.

For 5'-3'=aaatattgcatctggctgga (SEQ ID NO: 823)

Rev 5'-3'=aagtccaaaagtaactatgtaatctt (SEQ ID NO: 824)

New Rev 5'-3'=aatgacaagagtaaactcac (SEQ ID NO: 825)to exclude poly T region

M279= DBY int12n, site d ((nominal, 418 bp)) C to T at position 93, Site within STS with 7 T homopolymer.

Group I

For 5'-3'=aaatattgcatctggctgga (SEQ ID NO: 827)

Rev 5'-3'=aagtccaaaagtaactatgtaatctt (SEQ ID NO: 828)

New Rev 5'-3'=aatgacaagagtaaactcac (SEQ ID NO: 829) to exclude poly T region

#### **M280 revised** B9.36 c (386 bp) STS **G to A** at position 280

Group VI

newFor 5'-3' = ccagtcagcagtacaaaagttg (SEQ ID NO: 831) newRev 5'-3' = gcatttctttgattatagaagcaa (SEQ ID NO: 832)

M281 = G3.27f (393 bp) G to A at position 247.

Discovered while typing M123

tggtaaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGAT ACTAATTCATGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACACA GAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCAGCATCGCC TGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCAA AAAACTATGGGGGGAACAGGGAAGTCRGTTTAATAATACTGAGTTTGTGCA ACCTCAACTTTGCTTTATAGGAAAGCAAAATCTCAATATGATAAAGTTTTCTT CAACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaaga aaatctaattcgctg (SEO ID NO: 833)

For = tggtaaactctacttagttgccttt (SEQ ID NO: 834) Rev 5'-3' = cagcgaattagatttcttgc (SEQ ID NO: 835)

M282 = G3.27g (393 bp) A to G at position 316.

Group VI

tgg taaactctacttagttgcctttTGGAAATGAATAAATCAAGGTAGAAAAGCAATTGAGATACTAATTCATGCTCTCAGGGGAAAATCTGAATAAAGCTATCTTTTCTAACACAGAGCAAGTGACTCTCAAAGTCACAGTATCTGAACTAGCATATCAGCATCGCC

TGAATACCTAGAAATGCAAATTCCTGGGCAACACCAGAATCTAACAAAGCAA AAAACTATGGGGGAACAGGGAAGTCGGTTTAATAATACTGAGTTTGTGCAA CCTCAACTTTGCTTTATAGGAAAGCAAAATCTCAATATGATAARGTTTTCTTC AACAAAACTCTGAGATAACTATGTTGAGGGAAAGAAGTTGATCACATgcaagaa aatctaattcgctg (SEQ ID NO: 836)

For = tggtaaactctacttagttgccttt (SEQ ID NO: 837)

Rev 5'-3' = cagcgaattagattttcttgc (SEQ ID NO: 838)

M283 = DBY STS 09b (429 bp) A to G at position?

STS also contains M200.

For = ggcttacacttgcagactttg (SEQ ID NO: 840)

Rev: 5'-3'= ggagaaatgtacaagagtctaaacc (SEQ ID NO: 841)

M284 = EIF1AY STS34a, (399 bp nominal) del ACAA at position 105, STS has another marker, M306, GroupIX.

F 5'-3' = ggcagttttcatttaagcaga (SEQ ID NO: 843)

R 5'-3' = agcgaaactttcagcacttc (SEQ ID NO: 844)

M285 EIF1A\_Y STS12 (site d) (287 bp) G to C at position 70 (GroupVI)

newFor 5'-3'=ttatcctgagccgttgtccctg (SEQ ID NO: 846)

Rev 5'-3'=tgtagagacacggttgtaccct (SEQ ID NO: 847)

**M286** EIF1A\_Y STS12 (site e) (287 bp) **G to A** at position 129. (GroupVI)

newFor 5'-3'=ttatcctgagccgttgtccctg (SEQ ID NO: 849) Rev 5'-3'=tgtagagacacggttgtaccct (SEQ ID NO: 850)

M287 EIF1A\_Y STS12 (site f) (287 bp) A to T at position 100. This is one of 3 M201 related mutations.

(Group VI)

newFor 5'-3'=ttatcctgagccgttgtccctg (SEQ ID NO: 852) Rev 5'-3'=tgtagagacacggttgtaccct (SEQ ID NO: 853)

M289 = B9.36new d (386 bp) **G** to **A** at position 227 Group VI.

ccagtcagcagtacaaaagttgACAGCTTCAGCAAAATTGTAGCCTTGGTTAAAAACCACTG
TGGTAAGCACGAGGAAAAGTGATGACAAACTCCCCTGCACACTGGTTTGTGC
GGACAACCTAAAAAGGAGAAAAAAGCAGAAAGAGGTGTGGGTCAGAACTAA
TGGGCCAGATGTGAACTCAAAGATGTCTCTAGATGCTGTAACAGATGTAGGA
AGAGTGGAAARGCTCTATCTTCAAGTACGTGTCCTAAAAGAAAATGAGATTG
TGAATTTAAAAGTGGTATTCATAGAAAAGTACTCAAAATATGTGTAATTCAA
AAAACAAATATAGAGGGGTCCAGGAACAAGTGAAAAGACTCTttgcttctataatcaaa
gaaatgc (SEQ ID NO: 854)

For 5'-3' = ccagtcagcagtacaaaagttg (SEQ ID NO: 855) Rev 5'-3' = gcatttctttgattatagaagcaa (SEQ ID NO: 856)

M290 = B9.36new e (386 bp) G to A at position 343. Group III

ccagtcagcagtacaaaagttgACAGCTTCAGCAAAATTGTAGCCTTGGTTAAAACCACTG TGGTAAGCACGAGGAAAAGTGATGACAAACTCCCCTGCACACTGGTTTGTGC GGACAACCTAAAAAGGAGAAAAAGCAGAAAGAGGTGTGGGTCAGAACTAA TGGGCCAGATGTGAACTCAAAGATGTCTCTAGATGCTGTAACAGATGTAGGA AGAGTGGAAAGGCTCTATCTTCAAGTACGTGTCCTAAAAGAAAATGAGATTG TGAATTTAAAAGTGGTATTCATAGAAAAGTACTCAAAATATGTGTAATTCAA AAAACAAATATAGAGGGGTCCAYGAACAAGTGAAAAGACTCTttgcttctataatcaaa gaaatgc (SEQ ID NO: 857)

newFor 5'-3' = ccagtcagcagtacaaaagttg (SEQ ID NO: 858)

newRev 5'-3' = gcatttctttgattatagaagcaa (SEQ ID NO: 859)

**M291** = EIF1AY STS16, (480 bp) **A to G**, at position 358, (Group III)

cggagtctggctttgttggcCAGGTTGGAGTGCAGTGGCATGATCTCGGCTCAGGGCAATGTCCGTCTCCTGGACTCAAGCAGTTCTCCTGCCTCAGCCTCCCAGTAGCTGGGATTAGAGGTGTGTGACACCATGCCCGGCTAATTTTTGTATTTTTAGTAGAGATGGGGTTTCACCATGTTGGCCAGGCTGGTCTCGAACTCCTGACCTCAGGTAATGCACCCGCCTCGGCCTCCCAAAGTGGTGGGATTATAGGCGTGAGTAACCATGCCTGGCCTTTCACTCTTATTTTCTAAGAACTTTAGAATAATCACCGAGATATTCTAAAGTAAACAGGAATTTTTAATGGTTAAGCTRTTATTTGTCTTTTGTCATTTCTGAGTTTAGGGATAGTGAAGAGTTAGGCCTCATGTGTGAGAGACTGATGTAGCATTATATTTGAAATGTTATTTGTCTTTTGTCATTTCTGTCATTTCTAGCATTATATAGTGTATATTTTGAAATGTGCCCCgtgatgttcaaaagt(SEQ ID NO: 860)

For = cggagtctggctttgttggc (SEQ ID NO: 861) Rev 5'-3' = acttttgaacatcacggtgg (SEQ ID NO: 862)

M292 = EIF1AY STS19, (556 bp) A to G, at position 373.

Group III

Original F 5'-3' = tttaacaaatgtggaccaaga (SEQ ID NO: 864)

Rev 5'-3' = acttttgaacatcacggtgg (SEQ ID NO: 865)

M293 = EIF1AY STS20a, (507bp) T to G, at position 299.

Group III. STS also contains M294

F 5'-3' = catggtccaagcaatttatttttg (SEQ ID NO: 867)

#### R 5'-3' = gctggctaatacttccacagag (SEQ ID NO: 868)

M294 = EIF1AY STS20b, (507bp) C to T, at position 305

F 5'-3' = catggtccaagcaatttatttttg (SEQ ID NO: 870)

R 5'-3' = getggetaatacttccacagag (SEQ ID NO: 871)

M295 = EIF1AY STS20c, (507bp) T to C, at position 411,

(Group VIII). STS also contains M294 mutation

F 5'-3' = catggtccaagcaatttatttttg (SEQ ID NO: 873)

R 5'-3' = gctggctaatacttccacagag (SEQ ID NO: 874)

**M296** = EIF1AY STS21=STS20d, (536 bp) **C** to **T**, at position 165, (Group VIII)

F 5'-3' = gattgggggaaatagttttagg (SEQ ID NO: 876)

R 5'-3' = cactatgtgtggactaagccag (SEQ ID NO: 877)

**M297** = EIF1AY STS24, (506 bp) **A to G**, at position 303, (Group VII)

TtggttggtctacgggactATCAGGTAAAAATAACATTTAAAGTTGTGGTATGTCTGTGT TTAAGCAGTTGTTAATGTTTGGAAGGTAACTATACTAGCATCTTTGACCCATT CCAGCCCAGGTTGCTTTCTCACCATTCTGCCTGCCATCATCATTTATTAAGGG CCAGTTGTATTTCAGACTATAGTATTTTTCAAATTTGACATAATTCTCACTGAT AGTAAATGGTACATATTTTTGTGGAAAGACATAAAGTTTTTAATTCTTTGT TTTCATTGTTAATATAATGTGCAGTAAAT**R**TTTTCTTGCAGGCTTGGGCAAGT ACTGTAGACCATCTGTCCTCATCCATTTAAAGGCCAATGGTGTTTCAGGCATT CAGCTAGGTATTTCAGACATTGTAGTTCCCAAATGCCGGTCTGTTAAATAGTA TTGGTGCAGGCTGAATTTCAGTGCTCTGAAGTCAAATTAGAAGATACATAGT Tacgatgttttcatggagca (SEQ ID NO: 878)

F 5'-3' = ttggttggtctacgggact (SEQ ID NO: 879)

R 5'-3' = tgctccatgaaaaacatcgt\_(SEQ ID NO: 880)

M298 = EIFIA STS 27 (445 bp) G to A at position 230,

Group II

Original F 5'-3' = aaataccattttcataatttcctt (SEQ ID NO: 882) Original R 5'-3' = tgagatcctgcacagcaaga (SEQ ID NO: 883)

M299 = EIF1AY STS29, (483 bp) **T** to **G**, at position 127,

Group I

F 5'-3' = cggacttggtctgtgcttttc (SEQ ID NO: 885)

R 5'-3' = tgatctgtcacaatggcagt (SEQ ID NO: 886)

M300 = EIF1AY STS31, (500 bp) G to A at position 153, STS also contains M301, Group III

F 5'-3' = caggcaggtctactttcaatct (SEQ ID NO: 888) R 5'-3' = gtcaaacactgcaattcaaaac (SEQ ID NO: 889)

M301 = EIFIA STS 31 (500 bp) A to C at position 340bp. (Group III) STS also contains M300, a Group VII marker

F 5'-3' = caggcaggtctactttcaatct (SEQ ID NO: 891) R 5'-3' = gtcaaacactgcaattcaaaac (SEQ ID NO: 892)

M302 = EIFIA STS 32a (527bp) A to G at position 230 (Group VII)

F 5'-3' = caaagtgctgggattacagg (SEQ ID NO: 894)

R 5'-3' = cttctagcttcatctgcattgt (SEQ ID NO: 895)

M303 = EIFIA STS 32b (527bp) **G** to **C** at position 352, (Group X)

CaaagtgctgggattacaggCGCGAGCCACCGTGCCTGGCCTAGAAAAGTGTATTACCT
TTTTAACATCATTATTCTTTACTCCATTTTTAGTTTTGAATTGCAGTGTTTGAC
CTTAAAAGTTTTATATTACAATTTTTTTAATTAGTCTTTTATTTTTTCCAAGAG
ACTTCTAATTAAAAGGGAATAGTAAATAAAAGCACTGTGCTTGCCTTTTGTGC
TTTTATTAAAGTGAAATCTCTACAATCTTTCCTAAGCTGTTAATCACTGTTTAC
TAATGAACATAAACCACTTCCTAATTATTCAGACTCAAGAATTTTTTTCTAGA
GGGTATTGGGGTAGGCAAAGAAAASCAGGAGAGTTTGTAACAAACAGTATG
TGGGATTTTTTAGATGTTCAATTTGAAAGTAACTTGTGAAACAACTGGTG
ATATTTTGGTATAAGACGTTTTGAAAGTTATTTGTTTATTTCTAAGGATAACA
AAGCTGATGTAATTTAAAGTacaatgcagatgaagctagaag (SEQ ID NO: 896)

F 5'-3' = caaagtgctgggattacagg (SEQ ID NO: 897)

R 5'-3' = cttctagcttcatctgcattgt (SEQ ID NO: 898)

#### M304 = EIFIA STS 32c (527bp) A to C at position 421

F 5'-3' = caaagtgctgggattacagg (SEQ ID NO: 900)

R 5'-3' = cttctagcttcatctgcattgt (SEQ ID NO: 901)

#### M305 = EIFIA STS 33 (545 bp) C to T at position 331

AacttgtgaaacaactggtgatATTTTGGTATAAGACGTTTTGAAAGTTATTTGTTTATTTC
TAAGGATAACAAAGCTGATGTAATTTTAAAGTACAATGCAGATGAAGCTAGA
AGCCTGAAGGCATATGGCGAGCTTCCAGAACATGGTAAGATCAAAATGATTT
TATCTCCTCATTATTTGATATTAATGTTTGTTGGTATTTAGGTGAAGGTATTTC
CGTAGAACTCTTGTTTTACATACTGTTTTAGTGTATACTTAAAAATTTGTTATA
AGTAGTCTTGCCTATACTTCAGTTTACTTATGATACTTTGGAAAAGATATTAA
TAAYTGGAAATCTCTAATAAAAAACGTTATGAAACTTGAAAGTAGAAGTCTCTA
ATAAAGAGATTATGAATTATGAAAGTTCCTTTAGTGACAACTTTTATAAATTCA
TAAGCTCTGGATTTGTATATAAAGATCTGTCAAAGAAAATACGTTTTTTATAGTG
TTTTTCTAAACAGTTCTCAAGACTGGCAGTTTTCATTTaagcagaggcaacaaatgtaat
(SEO ID NO: 902)

F 5'-3' = aacttgtgaaacaactggtgat (SEQ ID NO: 903)

R 5'-3' = attacatttgttgcctctgctt (SEQ ID NO: 904)

M306 = EIFIA STS 34b (399 bp) C to A at position 231. Group IX. STS also contains M284, a Group VI marker.

F 5'-3' = ggcagttttcatttaagcaga (SEQ ID NO: 906)

R 5'-3' = agcgaaactttcagcacttc (SEQ ID NO: 907)

# **M307** = EIFIA STS 35 (500 bp) **G to A** at position 282 (Group VI)

F 5'-3' = ttattggcatttcaggaagtg (SEQ ID NO: 909)

R 5'-3' = gggtgaggcaggaaaatagc (SEQ ID NO: 910)

## **M308** = EIFIA STS 37a (444 bp) **T to C** at position 70 (Group I)

F 5'-3' = aaactttacagtcctttgggata (SEQ ID NO: 912)

R 5'-3' = tetcaagagaggaagaaaaate (SEQ ID NO: 913)

## **M309** = EIFIA STS 37b (444 bp) **A to G** at position 200 (Group II)

F 5'-3' = aaactttacagtcctttgggata (SEQ ID NO: 915)

R 5'-3' = tetcaagagaggaagaaaaate (SEQ ID NO: 916)

## M310 = EIFIA STS 37c (444 bp) C to T at position 352 (Group III)

F 5'-3' = aaactttacagtcctttgggata (SEQ ID NO: 918)

R 5'-3' = teteaagagaggaagaaaaate (SEQ ID NO: 919)

## M311 = EIFIA STS 39 (460 bp) G to T at position 304 (Group X)

F 5'-3' = cgagaacagcctaaccaaca (SEQ ID NO: 921)

R 5'-3' = gggtgtgatagatgaagcagag (SEQ ID NO: 922)

#### M312 = EIF1AY STS40a, A to T at position 49,

(Group VII)

AAATGCATTGTTGTTGATCCCAGGAAATCTGAGACAGGTCTCAGTTAATTT acaaagttgattttgccaaagt (SEQ ID NO: 923)

F 5'-3' = gtttccagactgttcagaggag (SEQ ID NO: 924)

R 5'-3' = actttggcaaaatcaactttgt (SEQ ID NO: 925)

#### M313 = EIFIA STS 40b Homopolymer 9T's to 10T's at position 288

For 5'-3' = gtttccagactgttcagaggag (SEQ ID NO: 927)

Rev 5'-3' = actttggcaaaatcaactttgt (SEQ ID NO: 928)

**M314** = EIFIA STS 40c (623 bp) **A to C** at position 419.

(Group VI)

F 5'-3' = gtttccagactgttcagagg (SEQ ID NO: 930)

R 5'-3' = aaggetaacaagatgecete (SEQ ID NO: 931)

M315 = EIFIA STS 41 (512 bp) A to C at position 395 STS also contains M314 GttcttgtgatcccaggaaatCTGAGACAGGTCTCAGTTAATTTACAAAGTTGATTTTGCC AAAGTTGAGGACGCACCCATGACACAGCCTCGGGAAGCCCTGAGGACATGT ACCCAAGGTGTTTGGGGCACAGCTTGGTTTACTACATCTTCAGGGAGACATG AGACATCAATCAATATATGTGAAAAGAACGTTGGTTCAGTTTGGAAAGGAG GGCATCTTGTTAGCCTTTCTAAAGGAGGCAGTCAGCTATGCATCTAACTCAAT GAGCGAAAGGATAACTTTTGAATAGAATGGGAGGCCGGTTTGTCTTAAGCAG TTTCCACCTTGAGTTTTCATAGTAATTTTGGGGGGCCAAAGATATTTTCGTTTC ACATTCTAATATTTTCTTCMTGTACCTCCCTTTGGGGGACCCTGAGCCAGAGGT

# TTTTTGGGGGATTAAACAGAATTGGCATTTACTTCATGTTGCAATAACCAAAA GCATAAATAttttgttgtagattaagggcaa (SEQ ID NO: 932)

F 5'-3' = gttcttgtgatcccaggaaat (SEQ ID NO: 933)

R 5'-3' = ttgcccttaatctacaacaaaa (SEQ ID NO: 934)

## M316 = EIFIA STS 42 (512 bp nominal) 5T's to 6T's at position 201 Group V

F 5'-3' = aattggcatttacttcatgttgc (SEQ ID NO: 936)

R 5'-3' = catgtccttacttccttttgtg (SEQ ID NO: 937)

# M317 = EIFIA STS 44 (523 bp nominal) –2bp Deletion of GA at position 400 (Group VIII)

F 5'-3' = tggttctacagttgggattttg (SEQ ID NO: 939)

R 5'-3' =ccttaataaccgaggcacaa (SEQ ID NO: 940)

#### M318 = EIF1AY STS20d, T to C, at position 353 Group VI

F 5'-3' = catggtccaagcaatttatttttg (SEQ ID NO: 942) Rev 5'-3' = gctggctaatacttccacagag (SEQ ID NO: 943)

#### M319 = UTY1 exon 14b, T to A at position 124. Group VI

GtaaaactcagatatatacatccatgAAATATACACAGAAACTATAAATTAGCATTAATATC CTCTAAAATGATACTGTAGTAAAAGAAAATATTCTCAAACTGTTGGTAAATTTTA GAGAAAAWAAAAATATTATACATACTTGCTGCATTAAGACAAACTGACTTTC TAACTGTTCCAGCTGATGCTTCTGTGCTGGATTTAAATTATCTCTATTTGCTCG CAGTTGTTCCAAGTGCTAGAAGAAAAAGAGATTAATAATCAAAGTTTAATC TAAAATTTAAGACAATATAAGGCAACTCCTCACTAAAAAGACTACACAGAAC CTTTGCAGGATGAAAGACAGTGATTCCTAATGAACgttaagatagtgattcttttttttt(SEQ ID NO: 944)

F 5'-3' = gtaaaactcagatatatacatcccatg (SEQ ID NO: 945) Rev 5'-3': aaaaaaaaagaatcactatcttaacg (SEQ ID NO: 946)

M320 = DBY STS08, (444 bp) T to G at position 60 Group VI

tgaggtggaatgtatccatataccaattaatatttttttaaaaaaatccttgaa atca (SEQ ID NO: 947)

For 5'-3'=tgaggtggaatgtatcagtatacc (SEQ ID NO: 948) Rev 5'-3'=tgatttcaaggatttgttagtctt (SEQ ID NO: 949)

M321 = DBY STS08, (444 bp) C to T at position 171 group VI

For 5'-3'=tgaggtggaatgtatcagtatacc (SEQ ID NO: 951) Rev 5'-3'=tgatttcaaggatttgttagtctt (SEQ ID NO: 952)

#### Footnote:

STS sequences (one strand only) for polymorphic Y sequences.

**Primer regions** = **lower case**; Reverse compliment made to generate 5'-3' Reverse PCR primer sequence for complimentary strand.

IUB code defines polymorphic site

R = A or G (puRine)

Y = C or T (pYrimidine)

K = G or T (Keto)

M = A or C (aMino)

S = G or C (Strong-3H bonds)

W = A or T (Weak-2H bonds)

H = A, C or T

Markers M1, M29, M40, M46, M130, M167, M176, M177, M222, M236, M288 are unassigned in TABLE 1.